Human
Genetics
Commission

Our genes, ourselves:
towards appropriate genetic testing

Third Annual Report of the Human Genetics Commission

2003

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Chair's introduction

It is my pleasure as I write this introduction to the Human Genetics Commission's third annual report to reflect on a year in which we have seen some significant changes in the Commission and in the wider arena that we inhabit.

We took the decision to slightly delay this report so that we could cover the publication of the Government's Genetics White Paper – "*Our inheritance, our future: realising the potential of genetics in the NHS*". The much anticipated White Paper has made a number of important announcements about how the Government intends to meet the challenges and opportunities presented by the advances in human genetics. This is being matched by major investment in training of NHS staff and equipping laboratories to improve the existing small and dedicated NHS clinical genetics laboratories.

The Commission was particularly pleased to see how the Government has responded to recommendations made in our 2002 report on personal genetic information. The White Paper, and the accompanying open letter from John Reid, the Secretary of State for Health, demonstrated that the Government truly is listening to our advice and, in turn, to the views that have been put to us in our consultations on human genetics.

Our work has clearly shown that there is a lingering unease about the possible uses that genetic information could be put to outside of the doctors surgery. We know that some of these fears are likely to be misplaced, but we do feel that there are important steps that can be taken – by Government and by other organisations – to develop a sense of trust.

We therefore warmly welcomed the decision to introduce an offence of non-consensual testing of human DNA. We want to consider the detail of the draft legislation to make sure that it addresses the main harm, namely the use of genetic testing in an intrusive and exploitative manner. We are particularly anxious to protect the well being of children and we are conscious that inconsiderate or reckless testing could greatly damage family relations. Yet, we do not want to interfere with rightful uses of testing.

One of the main themes running throughout our work this year has been how to regulate private DNA testing services – whether for medical or lifestyle purposes – so that they do not prey on people's ignorance or fears about health or infidelity. This was one of the conclusions from our report published in April 2003 called "*Genes direct: ensuring the effective oversight of genetic tests supplied directly to the public*". We originally saw this as simply building on the work on personal genetic information. But this turned out to be a fascinating and complex review which raised important issues around people's rights to access their own genetic information balanced against the fact that some tests can give misleading health information that overstates the role of genetics.

It is a particular source of pride that from the outset the Commission has been committed to openness in our methods of working – with public meetings, our agenda and minutes available on the website and the Consultative Panel of over 100 members of the public who are affected by genetic disorders. We tried some interesting methods of public consultation and found quite clearly that the public strongly supported the idea of protecting those they saw as vulnerable – the young, elderly and 'worried well' – and real need for people to be properly and independently informed before take such tests. We believe that the cornerstone of any decision to take a test needs to remain the provision of adequate information so that people can make the difficult choice themselves. The

NHS should be the main source of such advice and support, and we hope that the new investment will improve awareness of what genetic testing can and cannot tell us. There is also an important role for other health professionals such as pharmacists and complementary health practitioners. But inevitably such groups need to be able to distinguish their professional duties from their commercial interests.

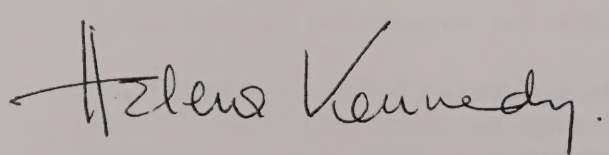
We will continue to be alert to individual circumstances where advertisements or printed reports appear to us to be over-stating the case for testing or for products related to genetic tests. We have been very pleased with the response to our concerns from bodies such as the Independent Television Commission and the Advertising Standards Authority. We have also been aided tremendously by the hard work of colleagues in the HGC's independent Press Office who have helped to forge some very effective links with the print and broadcast media.

In 2003 we reached the end of first 3-year 'term of office' and there have been a number of changes to our membership. We were pleased to welcome several new faces – Brenda Almond, Celia Brazell, Alastair Kent, Christine Patch and Peter Sayers – who I am sure will maintain the HGC's reputation. These changes gave us an opportunity to review how we work. We wanted to be able to keep track of all of the topics that had come onto our radar, as well as take forward important new work. We have set up a new, more flexible structure, focussing the main business of the Commission in task-orientated Working Groups, particularly on the use of genetic information in preparing for a family and in pregnancy. To keep a watching brief on some of the other key issues such as genetic discrimination, gene patents, genetic research, public involvement and horizon scanning, we have set up informal Monitoring Groups. For each of these we have identified Members to lead on each issue and make sure that these topics remain high on our agenda and that the Commission can successfully participate in wider discussions across Government and in external organisations.

However, we were all sorry to say goodbye to some of the original members of the Commission, especially Ruth Evans, John Polkinghorne and Gill Samuels. Our thanks are also due to Richard Pitts who returned to the Office of Science and Technology, to be replaced by Sophie Taysom as part of the Secretariat. I am sure that all of the Commission would like to thank our departing colleagues for their hard work and good humour which makes the Commission such a pleasure.

I want to make particular mention of Alexander McCall Smith who has been my most wonderful Vice-Chair since the creation of the Commission. His wisdom has been invaluable. However, amongst his considerable talents Sandy is a writer and the huge international success of his novels means that he is having to reduce his many public commitments. We are bereft that he will be stepping down shortly but thrilled at his success – no one deserves it more.

I hope that you find this annual report useful and informative and I hope that we can count upon you to be part of our future work and consultations.



Helena Kennedy
Chair, Human Genetics Commission

Contents

Our meetings this year

"In conducting its work, the HGC has been a model of openness and transparency. It has sought innovative ways of engaging the general public."
Government White Paper on Genetics

Continuing our commitment to holding our main meetings in public we again visited a number of places this year. The papers and minutes of our meetings and reports of the preceding information-gathering sessions are published on our website (www.hgc.gov.uk). Details of the sessions are included at Annex B.

"Holding open plenary meetings is one of the best public education strategies of the Commission." Plenary attendee

September 2002 – Belfast

We held our plenary meeting in Belfast on 11 September where we were warmly welcomed by Lord Alderdice and Barbre de Bruin of the Northern Ireland Assembly. Our thanks go to Patrick Morrison and his colleagues and those at the W5 centre for organising the visit. Our discussions focussed mainly on progress on the forthcoming Department of Health's Green (later White) Paper on Genetics and how HGC's work concerning genetics and reproductive choice might be taken forward. Members also noted various inquiries concerned with the storage and use of human tissue, ongoing work in the area of gene patenting, and recent developments in stem cell research and therapy. The meeting concluded with a discussion with the audience on issues related to our review of genetic testing services supplied direct to the public.

The day before we held an information-gathering session on pharmacogenetics, the study of how genetic variation can affect a person's response to a drug. The four presenters gave us an understanding of what pharmacogenetics is, what it can do now and might be able to do in the future, and the wider social and ethical implications that might arise. We heard about the potential health benefits that pharmacogenetics might bring, and some of the social and ethical issues which it raised, concluding that the situation was a complex one, and advances in this field needed to be monitored, as did those in related areas such as information technology.

November 2002 – London

In November in London, our discussion centred on the review of direct access genetic testing services and such concerns as the impact on individuals of predictive genetic tests without appropriate support or counselling. Consultation had shown that most people would prefer to approach their GP for such tests and we agreed that most genetic tests should not be offered direct to the public. Our recommendations would need to reflect the views put to us by the public, consumer groups, professional bodies and commercial organisation. We also discussed the UK Biobank project and agreed to change our ways of working, setting up more flexible and issue specific groups to lead on pieces of work.

The discussion on the UK Biobank large genetic database project was prompted by a request from the House of Commons Science and Technology Committee for a memorandum on the subject. It followed an information-gathering meeting the day before at which five presenters covered the

scientific rationale behind Biobank, oversight and governance issues, concerns about consent/ confidentiality and data security, and commercial access to Biobank. The subsequent discussion included further topics such as police access, intellectual property and patenting rights, feedback and public consultation.

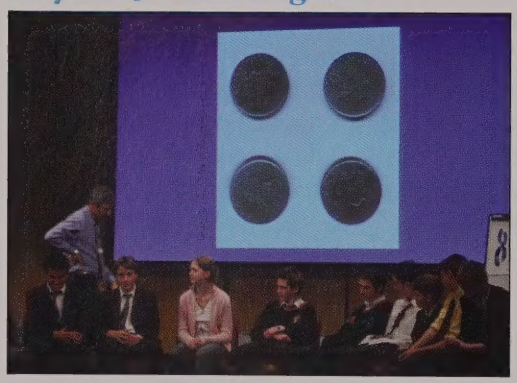
"Thought it an excellent meeting – well chaired and truly participatory." Plenary attendee

February 2003 – London

We met again in London on 5 February, where we welcomed our five new Members and we agreed the report and recommendations for our review of genetic testing services supplied direct to the public. Members also agreed a new structure for working, with Lead Members monitoring areas such as genetics and insurance, genetic research, gene patenting, horizon-scanning and public involvement, and supported by others who had an interest and possibly a different perspective.

We also took the opportunity to hold informal discussions around aspects of genetics and reproduction, and around our new work programme for 2003-2005. The first included presentations on issues arising from use of assisted reproductive technology (ART), and a couple screening model used in relation to antenatal cystic fibrosis testing. The discussions on the work programme covered topics such as genetic information, genetic testing and services, and communication with the public.

May 2003 – Birmingham



We went to the ICC in Birmingham on 6 May, where we and a number of local school children were entertained by John Burn's enjoyable and thought-provoking explanation of some of the basic principles of clinical genetics. We also took advantage of the European Society of Human Genetic's (ESHG) annual meeting which was held at the same time to hear about communicating genetics in an European context. We learnt about the situations in Italy and the Netherlands and about ongoing work with health professionals in the UK. Our thanks go particularly to John and to Anna Lane, for a successful event.

The following day our main item of business was how to progress our work on genetics and reproduction. We discussed draft terms of reference for the new Working Group on Genetics and Reproductive Decision-Making, and explored some of the issues which should be covered. The importance of liaising with organisations such as the National Screening Committee and the Human Embryology and Fertilisation Authority was highlighted. Lead Members from each of the Monitoring Groups updated Members on relevant activities, and Members agreed that monitoring development of the National Police DNA Database should be added to HGC's work plan 2003-2005.

"I think it is worthwhile as it gives people a glimpse of how decisions are reached and what types of people are involved in making them. I think it improves knowledge of what the HGC is doing and encourages people to get involved in consultations etc." Plenary attendee

Genes direct

This year we carried out what proved to be a fascinating and complex review of the possible developments in testing, of public attitudes to genetic tests and of the current and planned regulatory framework. This resulted in our report *'Genes direct – Ensuring the effective oversight of genetic tests supplied directly to the public.'* published in April 2003.

Our review showed that genetic tests supplied direct to the public raise some serious issues and we concluded that there are some potential harms from the sort of genetic tests that may be marketed directly to the public. A robust but flexible regulatory system is therefore crucial.

Safeguards are needed to prevent abuses and to subject direct genetic testing services to appropriate oversight. But we did not feel there should be statutory prohibition of direct genetic tests. Our report reflected our finding that the public trust and rely on the NHS for access to predictive genetic tests. We felt strongly that there should be a well-funded NHS genetics service supported by a genetically literate primary care work force, which can properly manage and allow access to new predictive genetic tests that are being developed. This would enable predictive genetic testing to be retained within a well-respected model of continuing healthcare. In view of this, we concluded that:

- most genetic tests that provide predictive health information should not be offered as direct genetic tests
- predictive genetic tests that are done at home should be discouraged until there are proper safeguards

We decided on a varied programme of consultation starting with a consultation document that received a good deal of interest and began a public debate. We took evidence from a broad range of interests, including regulators, consumer groups, professional bodies and commercial organisations. We also wanted to find out what potential consumers felt and arranged a series of focus groups, discussion meetings and an opinion survey via the Internet. The various responses and reports are available on our website (www.hgc.gov.uk).

- **Consultation responses** – in July 2002 we published a consultation document, and received 65 responses.
- **Evidence-gathering meetings** – while the consultation was underway, we held a series of evidence-gathering meetings with key groups and individuals to hear views first-hand.
- **Consultative Panel** – the HGC Consultative Panel discussed this at their July meeting (see page 8).
- **Wider consultation** – recognising that the market for direct genetic testing services will be largely driven by consumer demand, we wanted to hear from the wider public so conducted a series of focus groups, an internet-based survey and a series of public discussion meetings.

We did not make detailed recommendations about implementation as it is not our role to provide specific details of the wider regulatory framework. We took careful note of plans for the Medicines and Healthcare Products Regulatory Agency, the Council for the Regulation of Healthcare Professionals and a possible new Human Tissue Authority. And we recognised an important role for industry and professional self-regulatory bodies.

It is also one thing to regulate the UK market, but we noted the difficulties in regulation of the Internet. We felt that consumer awareness and common sense will be important safeguards against the possibility of widespread uptake of direct genetic tests. We recommended funding for consumer bodies to provide impartial advice that is available from trusted sources like NHS Direct or ourselves.

Finally, we do not believe that this is the final word on this matter. Because of the changes in the regulatory framework and the development of the market we felt that we should host a meeting of all relevant groups in 12-18 months to consider progress and to highlight any areas that require further consideration.

Genes direct was published in May 2003 and attracted a number of comments.

'Genetic tests must not go on public sale, Government told'
The Times 10/4/03

'DIY tests under fire'
The Sun 10/4/03

'GeneWatch UK criticised today's new proposals on the regulation of human genetic tests as "a triumph of spin over substance"'
GeneWatch UK press release 9/4/03

'Genetic home-testing kits "need strict monitoring"'
Independent 10/4/03

'Genetic test sales face restrictions'
The Scotsman 10/4/03

'New recommendations on over the counter testing kits – a missed opportunity by the Human Genetics Commission'
Sciona press release 9/4/03

'Industry wholly supports the caution with which the HGC have approached this complicated and emotive issue.'
BIVDA press release 9/4/03

'Commission warns against selling genetic tests direct to the public'
British Medical Journal 12/4/03

'Commission wants stricter controls on genetic test kits'
Press Association 9/4/03

Government response to Inside Information

In June we welcomed the publication of the White Paper *"Our inheritance, our future: realising the potential of genetics in the NHS"*. The Genetics White Paper set out the Government's strategic vision for genetics and healthcare and gave a plan of action and investment aimed at making the NHS a world leader in genetics based healthcare. Whilst we welcomed the extra investment in NHS genetics over the past two years – in the White Paper and previous speeches by the Secretary of State for Health – we also felt that it may be a drop in the ocean if genetics is truly to become a mainstream clinical tool.

The extra investment is aimed both at the specialised clinical genetic service in England, which includes laboratory modernisation IT systems and staff. This is supported by improved education and training in mainstream NHS services that will benefit from genetic advances. It is important to also note that the developments in genetic services and training in England are being matched as part of similar reviews completed in Wales and Northern Ireland and planned for this year in Scotland. The Genetic Services Subgroup has decided that one role for HGC is to consider the overall picture of genetic services in the four home countries (see page 14)

The White Paper dealt with some of the legal and ethical concerns raised by advances in genetics. It included commitments to a sound regulatory framework and to develop policy in an open and transparent manner. There is investment in the public understanding of genetics. We were very pleased to see that the Commission merited a number of extremely supportive comments, particularly our role in anticipating and addressing issues of public concern.

The White Paper forms the main element of the Government's response to Inside Information and is supplemented by an open letter from the Secretary of State for Health to Baroness Kennedy (Annex D). We are pleased that Ministers felt our conclusions and the ethical principles we set out influenced the Government's thinking in this area.

"We would like to thank you once again for your thoughtful and comprehensive report. We believe that it has made an important contribution to the development of appropriate safeguards to allow society to take advantages of the advances in genetic knowledge."
Secretary of State for Health

"We warmly welcome HGC's clearly-argued overarching principles, especially the balancing of respect for individual rights (for privacy, confidentiality and non-discrimination) with the need for "genetic solidarity and altruism" so that genetic knowledge can be shared to help others and society. These two principles form a valuable basis for developing future Government policy on genetics."
Government White Paper on Genetics

Some of recommendations in *Genes direct* followed directly from our earlier concerns about non-consensual DNA testing that we identified in our earlier report *Inside Information*. We were pleased by the announcement that the Government will introduce a new offence of testing an individual's DNA without their knowledge or consent by means of a new Bill that will regulate the removal, storage and use of human organs and tissues. The new offence will not interfere with medical situations where consent is impossible to obtain, or DNA analysis by the police and courts.

"we will introduce a new offence of testing an individual's DNA without their knowledge or consent by means of a new Bill that will regulate the removal, storage and use of human organs and tissues."
Government White Paper on Genetics

The Government accepted that the prospect of 'genetic discrimination' is an extremely complex matter and will become increasingly relevant with advances in the use of predictive genetic information by the NHS and privately for occupational health purposes. Although this is not yet a widespread problem, the Government accepts the HGC's recommendation and will consider the evidence for unfair discrimination on the basis of a person's genetic characteristics and the appropriate means of addressing any concerns in this area. It is actively considering the most appropriate way to take this forward. The Government also asks the Human Genetics Commission, with others such as the Disability Rights Commission, to continue to monitor developments.

"A core ethical principle in the HGC report is that no one should be unfairly discriminated against on the basis of his or her genetic characteristics. The Government wholeheartedly endorses this principle." Government White Paper on Genetics

The White paper and Government response can be found on the DH website (www.dh.gov.uk/genetics/

New work for HGC from the Genetics White Paper

Profiling babies at birth

The White Paper included a request for the Commission with the National Screening Committee, to consider the case for profiling babies at birth and storing information about their genetic profile for future use. Whilst this attracted considerable publicity, we have noted that there are no plans to introduce any such service. The White Paper states that a more thorough knowledge about the meaning of genetic variations, together with public debate on many of the wider issues would be needed before any such a system could even be considered. We have therefore added this to our work plan alongside our main work on genetic testing and decisions about pregnancy.

Behavioural genetics

HGC noted and praised the Nuffield Council report in making recommendations about direct genetic testing for behavioural traits. In Genes Direct we stated that "we will continue to monitor developments in this area in the UK and elsewhere". In the White Paper, the Government echoes this and therefore asks HGC to monitor research into behavioural genetics and its possible applications and to advise Government of any further action that may be needed.

The Nuffield Council on Bioethics report in October 2002 extensively reviewed the field. It raises some concerns that genetic information about behaviour might be used to 'medicalise' normal behaviours, or to market certain genetic tests. It also raises concerns about 'eugenic' uses of such information as well as its misuse in the contexts of education, employment, insurance and criminal justice. The report includes some recommendations to be taken into account in considering the need for legislation to protect people against unfair genetic discrimination.

Genetics and Reproductive Decision Making

New and developing technologies associated with genetics and human reproduction, and their implications for society, have raised public concerns. In particular, the possible implications of genetic screening and testing for reproductive decision making have been flagged up as issues. In response, during the year, HGC began a new major piece of work on genetics and reproductive decision making.

In deciding how to move this work forward, and what key issues are to be addressed, in December we contacted about 90 organisations including various charities, professional organisations and religious groups who might have an interest in this topic. We also later wrote to our Consultative Panel asking for any suggestions they might have.

We had meetings in December 2002 and April 2003. The aim of these was to identify key issues in order to shape the work to come. Between these meetings, we also commissioned a piece of work on 'Choice, and the philosophy of choices applied to reproductive choices' by Heather Draper and Tom Sorell. Then, as part of the second scoping group meeting, we held an information gathering session on the topic of 'Genetics and Reproduction in the 21st Century: The myth of choice?'

At the May plenary meeting, HGC shifted its focus from scoping to working on this topic, hence the development of the working group on genetics and reproductive decision making. The group held its first meeting in June 2003.

A key aim of this project is to collate information, take evidence and consider past, current and future developments in genetics services related to reproduction within the current legal framework and in terms of the technology and public attitudes towards its use. This will build on earlier work conducted done via a joint working party between HGC and the Human Fertilisation and Embryology Authority on the topic of preimplantation genetic diagnosis. We will also continue to build on dialogue with a range of key stakeholders, HGC's consultative panel and the public. It will also involve working closely with, amongst others, the Human Fertilisation and Embryology Authority and the National Screening Committee.

The Working Group is co-chaired by Helena Kennedy and Martin Richards.

Genetics and Reproduction in the 21st Century: The myth of choice?

On the 8th April 2003, we held an information event in London on genetics and reproductive decision making. The meeting was attended by about 70 people, including members, members of HGC's consultative panel and invited guests.

'The best I have attended in terms of clarity of the presentations and response of the "audience".' Event attendee

We had four speakers present their views on aspects of this topic including:

- Bishop Harries of Oxford discussing 'Human responsibility and 'the myth of playing God'
- Dr Celia Roberts reporting on her findings from a project covering 'Genetic information and choice in the context of reproductive decision-making: findings from an ethnographic study of Preimplantation Genetic Diagnosis'
- Dr Anne Kerr discussing 'Reproductive genetics: putting "choice" in context'
- Professor Peter Braude raising issues around the question 'Should we choose our children?'

'Mostly, I feel "moral/ethical" issue have been "ducked" partially because they have been considered in earlier discussions.' Event attendee

These presentations were followed by a panel discussion where members of the audience had the opportunity to raise and discuss issues, or seek the views of the speakers on issues of genetics, reproduction and choice. Many aspects of these discussion where then taken on board as topics to be addressed when HGC considers genetics and reproductive decision making.

'these decisions should not be "left to the state" but must have "democratic" discussions and feedback. The HGC is doing this in a very open way and it is influencing other bodies to operate in a similar way.' Event attendee

Further details of this work including a report of the day can be found on HGC's website.

Consultative Panel

The Consultative Panel is made up of over 100 people with direct experience of living with genetic disorders, acts as a sounding board for our reports and recommendations, as well as giving us insight into their concerns about genetic issues. It has been an invaluable resource for us and a useful way of developing and checking our conclusions and recommendations. The Panel is also increasingly seen as a useful resource for others, such as other scientific bodies or research groups.

Panel Members have been involved in a number of areas of work. As well as the discussions at the July meeting (see opposite), we sent Members our draft recommendations from the Genes direct report for comments. A number of Members also took part in the Democs meetings that were run as part of the public consultation on this issue.

Members also contributed to the work of other organisations, for example the Royal Society's People's Science Summit 'Genetic Testing: which way forward?' in March 2003. The Summit, provided an opportunity for general members of the public, scientists and representatives of other groups to discuss and make joint recommendations concerning government policy on genetic testing. A report of the discussions and the recommendations which resulted from the day's proceedings can be found on the Royal Society's website (www.royalsoc.ac.uk).

A number of Panel Members also contributed to the process of drawing up the work plan. We asked Members what they saw as the most important issues we should be addressing in the future. Many agreed with the priorities we had suggested (see page 15) and their views helped us to decide how to order these priorities.

The first meeting of the HGC Consultative Panel was held on 16 July and was attended by around 40 members of the Panel. The agenda included introductions from Helena Kennedy and Ruth Evans and presentations from John Sulston, who gave an overview of the human genome project, and John Burn, who spoke about his work on cancer genetics. In the afternoon there were roundtable discussions on genetic testing services supplied direct to the public and genetics and reproductive choice, followed by a general feedback session.

We aimed for an informal meeting offering a chance for the Panel to meet each other and HGC Members, discuss a couple of issues in more detail and to ask any questions or make any suggestions they might have. There was no attempt to record the exact discussions at each table but a note was made of the general discussion afterwards (annex C).

Genetic Discrimination Monitoring Group

Following publication of our recommendations on genetics and insurance in 2001 and on genetic information in 2002, we decided to maintain a watching brief on developments.

Bill Albert has taken on the role as the Lead Member of HGC on genetic discrimination, in consultation with others on the Monitoring Group. The Group has collated a list of HGC's previous recommendations and, where possible, the Government's response to these.
The main focus is on:

- Developments that might be relevant to a long-term policy after the end of the moratorium on genetics and insurance (scheduled to end in 2006).
- How family history information is used by insurers and whether it is was being used as a simple surrogate for genetic test results during the insurance moratorium.
- Developments in the use of genetic test results in employment, particularly whether there was evidence of widespread use.

The Group has drawn together research work and discussion workshops on aspects of genetics and insurance, for example the workshops organised by the Genetic Interest Group with funding from the Association of British Insurers. The Group has noted with interest the work of the reformed Genetics and Insurance Committee, particularly in revisiting the criteria used to assess genetic tests for insurance uses.

The Lead Member has met with other groups such as the Disability Rights Commission, the Association of British Insurers and the Royal College of General Practitioners.

The role of the group is to oversee HGC activities relating to genetic non-discrimination, particularly in insurance and employment and monitor the work of other relevant bodies to ensure effective and efficient collaboration.

Members
Bill Albert (Lead)
Stephen Bain
Hilary Harris
Alastair Kent
Martin Richards
Peter Sayers

The Lead and Group members hold regular telephone conferences and bilateral meetings to identify work for HGC and to monitor the work of other key stakeholders and new research findings. The Lead Member and group has held discussion with outside bodies and where possible notes of such meetings are on the HGC website.

13 February 2003
21 May 2003

In June, the Group was pleased to see that the Government had accepted the HGC's main recommendation on the possible need for a specific legislation to prevent genetic discrimination. The Government has also reaffirmed HGC's role in monitoring developments and working with the Disability Rights Commission, the Health and Safety Commission and others.

"Although this is not yet a widespread problem, the Government accepts HGC's recommendation. We will consider the evidence for unfair discrimination against people on the basis of their genetic characteristics and the appropriate means of addressing any concerns in this area." Government White Paper.

Horizon-Scanning Monitoring Group

An important role of HGC is to provide Government with advice on advances in human genetics and their implications for healthcare as well as the broader social and ethical implications.

The Horizon Scanning group has continued its work by considering a number of important issues including:

- **Pharmacogenetics** – where people were tested for a likely response to a particular drug before being given that drug whether as part of a research study or to help with prescribing decisions. While still at an early stage, the potential use of pharmacogenetics raises a range of social, ethical, legal and economic issues. For this reason, the horizon scanning group helped organise an information gathering session on the topic of pharmacogenetics held in September 2002.
- **Research and UK Biobank** – in addition to considering strategic priorities for research, the group helped organise another information gathering session in November 2002, this time on UK Biobank. The UK Biobank project will be the world's biggest study of the role of nature and nurture in health and disease. Up to half a million participants aged between 45 and 69 years will be involved. This however raises a range of social and ethical issues which HGC looked at in this session. Details of this session and the session on pharmacogenetics can be found at **Annex B**.
- **Reproductive choice** – where it was noted that technology would have implications for prenatal genetic diagnosis, preimplantation genetic diagnosis and screening, as well as for treatments which would have an impact on choices available. This is now a major topic for HGC's work and the horizon scanning monitoring group have and will continue to play an important role in examining this topic.

The role of this group is to identify and monitor developments relating to horizon scanning, particularly the ethical, social and legal implications, and report these back to HGC to consider.

Members
Veronica van Heyningen (Lead)
Celia Brazell
John Harris
John Sulston

The horizon scanning group monitors developments in human genetics. As its work cross-cuts the work of other monitoring groups and working groups, it was decided that the best use of this group could be made through assisting HGC to reach a view on new developments rather than producing a single all-embracing report. In addition, there are a number of issues that have been or are being examined by other bodies such as the Nuffield Council on Bioethics. It is the role of this group to maintain a watch of publications in this area and report to the main Commission.

Intellectual Property and Genetics Monitoring Group

The issue of patents, intellectual property and genetics continues to raise concern in many national, regional and international forums. HGC has established an intellectual property and genetics group in order to monitor these concerns as well as European and international developments of relevance to the UK.

We have continued to keep watch on developments in the area of patents and genetics. This topic is also now a standing item on the plenary agenda. In contributing to the plenary meetings, during the year our focus has largely been on:

- Effects of gene patents – in the last year, reports on the effects of patenting in science have been released by the Royal Society, the Commission on Intellectual Property Rights and Nuffield Council on Bioethics. These have been of great interest to both this group as well as the Commission as a whole.
- Proper application of the law – in relation to striking a balance between private and public interest, it is increasingly important to analyse and monitor how the law is being applied.
- Competition and access to information – one significant public concern is the potential for patents on genetic tests to limit their availability. We have monitored, and will continue to track developments with respect to this.
- National, regional and international developments – the key role of the intellectual property and genetics group has been to monitor developments with respect to the legal, as well as social, economic and ethical implications of national, regional and international developments.

Our role is to identify and monitor developments relating to intellectual property and report these back to HGC to inform and consider.

Members
Hilary Newiss (Lead)
Brenda Almond
Celia Brazell
Alastair Kent
John Sulston

In February 2003, an Intellectual Property monitoring group was formed to build on the work done by other bodies in fostering debate, to monitor developments as well as publications by other bodies. The Lead and Group members have regular email contact to monitor the work of key stakeholders, new research findings as well as to begin to form views on a variety of topics. Since February, the main focus of this group has been on updating relevant sections of the HGC website.

Public Involvement Monitoring Group

Real public involvement underpins all of our work, a fact which was recognised by the Government in its recent White Paper.

We initially had a sub-group which has helped to develop HGC's overall approach, as set out in the Public Involvement Strategy. Now that these procedures are established, we wished to see public involvement treated as an integral part HGC's work on specific issues.

We therefore disbanded the Sub-group and established a Monitoring Group to maintain oversight of the main strands of public engagement work, such as:

- Public Involvement Strategy
- HGC's Consultative Panel
- HGC's Press Office
- Website

as well as keeping up to date with what others are doing.

The purpose of this group is to oversee HGC's activities relating to public involvement, in particular to promote debate and achieve effective representative dialogue with a wide cross-section of people.

Members
Geoff Watts (Lead)
Elizabeth Anionwu
John Burn
Peter Sayers

The Group met on 3 June when discussions focusing on the development of the HGC website. This is something we have agreed to look at as a priority and we want to move forwards on improving both the structure and the content of the site.

We have been extremely pleased with the way that our work in the vanguard of public involvement has been received by Government, other advisory bodies, learned societies and the wider public. We have been active in explaining and promoting our work and at the same time we have been receptive to innovative approaches to public engagement. Our consultations for the "Genes direct" report made use of two different procedures (DEMOCS and YouGov) and we were pleased with the results.

The Press Office carried out a 'perception audit' for us in January and February this year so we could get an idea of people's views on the Commission and whether or not we were achieving what we set out to do. The audit involved interviewing varying organisations and individuals, journalists and HGC Members past and present and we included a range of organisations, not just those we thought were likely to rate us in a positive light. This has given us a wealth of views, feedback and suggestions that will help us plan our future work and public involvement activities.

Research Databases Monitoring Group

Our Inside Information recognised the importance of genetic research and we wanted to ensure that people are willing and able to participate in it. We want to achieve a balance between the needs of researchers, while at the same time properly protecting people's genetic information and being very clear about how it will and will not be used.

Our report addressed general research issues like consent and feedback and confidentiality. It also covered the particular issues raised by large-scale research databases. We have, through our Lead Member, Martin Richards, and the Research Databases Monitoring Group, been closely following the progress of the UK Biobank project (www.biobank.ac.uk). This aims to collect samples from 500,000 people to help explore the relationship between genes, environment and disease.

In October 2002, the House of Commons Science and Technology Committee requested a memorandum from HGC on UK Biobank, as part of its enquiry into the work of the Medical Research Council. In order to raise and discuss relevant issues, HGC held an information-gathering session in November 2002. A report of the meeting and the Memorandum to the Committee can be found on the HGC website (www.hgc.gov.uk).

The Commons Science and Technology Committee's report[1] in March 2003 reflected many of HGC's points and the Government response[2], produced in June was followed by further specific responses to HGC in the Genetics White Paper.

As part of its ongoing work, the Monitoring Group has met with the Chief Executive of the UK Biobank and the Chair of the Funder's Interim Advisory Group and remains committed to a continued dialogue to help ensure the success of this important research tool.

The role of this group is to oversee HGC activities relating to genetic research and databases, particularly the ethical, social and legal implications of large projects such as the UK Biobank.

Members
Martin Richards (Lead)
Stephen Bain
Celia Brazell
John Burn
John Harris
Hilary Newiss
Veronica van Heyningen
Hilary Harris

The main areas of interest to the Monitoring Group include:

- ensuring consent that it is fully informed and covers questions like feedback and intellectual property
- ensuring strict confidentiality, by effective anonymisation, encryption and by controlling access by groups such as the police
- Maintaining public confidence, particularly ensuring that large research databases remain a trusted public resource
- Promoting realistic expectations of the pace of scientific and medical research and the role of partnerships between public and commercial research

We have held public meetings and informal liaison meetings with the Biobank funders. The Monitoring Group met the Funders on 18 June 2003.

1 House of Commons Science and Technology Committee. 24 March 2003. The Work of the Medical Research Council, Third Report of Session 2002-3. HC132

2 Department of Trade and Industry. June 2003 Government response Cm5834. HMSO

Genetic services

There have been two main themes for the HGC's Genetic Services Sub-group this year – direct genetic services and the proposed Green (later White) Paper on genetics.

The Groups consideration of 'over-the-counter' genetic testing services led to the setting up of a Working Group which consulted on the issues and produced the Genes Direct report. The Sub-group's chair, Philip Webb, also led on the work on direct genetic testing services, and the larger Sub-group was involved in finalising the consultation document and overseeing the arrangements for gathering wider evidence. It also considered some final follow-up around the service that was launched by Sciona Ltd. in 2001.

Our work on genetic services has been largely one of monitoring the major developments in NHS genetic services that followed from Alan Milburn's speech in April 2001 on genetics in the NHS. This led to major investment in NHS services, training of professionals and reviewing the arrangements for commissioning services. It also promised a Green Paper on genetics which was the subject of a conference in January 2002 at which HGC were present. There were also reviews of genetic services in Wales and Northern Ireland, and planned for this year in Scotland.

The Sub-group continued to take a close interest in the work of the Genetics Commissioning Advisory Group and for a Genetic Testing Network to improve co-ordination of NHS genetics laboratories in providing tests, especially for rare conditions.

In late 2002 the Sub-group was reviewed and it was decided that its role should change slightly and should provide a wider perspective of genetic services across the UK. This would in part be done by having a rolling programme of meetings in England, Scotland, Wales and Northern Ireland. This would enable HGC to better fulfil its remit of providing strategic advice on the healthcare aspects of human genetics.

As part of this, the Membership was reduced and the Sub-grouped thanked the several HGC members and co-optees, Bob Bestow and Heather Draper, for their valuable contributions.

Genetic Services Sub-group

(membership after February 2003)

Chair: Philip Webb

Members:

Elizabeth Anionwu
Hilary Harris
Alastair Kent
Christine Patch
Peter Sayers
John Sulston
Frances Flinter (co-optee)
Peter Harper (Wales CMO)
Patrick Morrison (NI CMO)
Stephen Singleton (England CMO)
Rosalind Skinner (Scotland CMO)

Remit: To keep under review and advise the Commission of new issues and developments in the following areas, including the routine exchange of information with other relevant bodies such as the National Screening Committee, the Genetics Commissioning Advisory Group and the Human Fertilisation and Embryology Authority:

- Strategic issues in the delivery of genetic services by the NHS and the private sector;
- Human genetic testing services supplied direct to the public;
- Significant new and evolving genetic tests and screening and associated technologies; and
- Codes of practice and guidance on the ethical, social and scientific aspects of human genetic testing services and their effectiveness.

Work plan 2003 to 2005

During the year, we drew up a 3 year plan of work from 2003 to 2005. This was discussed on a number of occasions throughout the year. We took stock of what we had achieved against the objectives set in previous work plans and identified priorities for the future.

Our key achievement during the year was the publication and launch of 'Genes Direct'.

As HGC does not work in isolation, we wanted to hear what others with an interest saw as priorities for our work. As part of this, we wrote to about 120 organisations, the Department of Health, other Government Departments and HGC's consultative panel. We have now taken these comments into account in our work plan. Most respondees were reasonably content with what we were suggesting. One major addition was the Ministerial request in the Genetics White Paper to work with the National Screening Committee to consider the case for genetically profiling babies at birth.

In addition to this work with the National Screening Committee, we identified the following as priorities for the following three years:

- To consider issues around genetics and discrimination, particularly in insurance, and have an active input into decisions about what should happen after the current moratorium;
- To review the ethical and social implications of new and developing genetic technologies for reproductive decision making. This will involve working closely with others such as the National Screening Committee and the Human Fertilisation and Embryology Authority.
- To conduct a short review of genetic relationship testing and services such as paternity testing;
- To further find ways to expand HGC's public involvement, such as through developing our website as an important resource in studying the ethical, legal and social implications of human genetics;
- And finally, to monitor issues across a range of HGC topics including research and databases, intellectual property, public involvement, genetic services and horizon scanning.

Much of the work on these topics is ongoing. For example, HGC's Discrimination Working Group has been involved in some discussions about insurance and what happens after the current moratorium, and the work on the ethical and social implications of technologies for reproductive decision making has begun.

This work, as with all HGC work, will of course adopt a UK perspective where we will take into account the legal and other difference between Scotland, Northern Ireland, England and Wales, and of the status of devolved and non-devolved matter.

Keeping in touch

Tell us what you think

We are always keen to hear what you think and comments are welcomed about

- the report we published on genetic tests supplied direct to the public
- the issues we have identified in our work plan, in particular on genetics and reproductive decison-making
- how we can involve people in our work
- or any of the issues in this report

How?

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Find out more

Website

To find out more about the HGC please visit our **website:** www.hgc.gov.uk

Details of HGC publications are given in Annex H

ANNEX A: Membership

The Human Genetics Commission

Chair
Baroness Helena Kennedy
Barrister and broadcaster

Vice-Chair
Professor Alexander McCall Smith
Professor of Medical Law, University of Edinburgh

Members
Dr Bill Albert
Chair of the Norfolk Coalition of Disabled People

Professor Brenda Almond (from January 2003)
Professor of Moral & Social Philosophy Hull University

Professor Elizabeth Anionwu
Professor of Nursing, Head of Mary Seacole Centre for Nursing Practice, Thames Valley University

Dr Stephen Bain
Reader in Diabetic Medicine at Birmingham University and Consultant Physician at Birmingham Heartlands Hospital NHS Trust

Dr Celia Brazell (from January 2003)
Director of Science and Technology GlaxoSmithKline

Professor John Burn
Professor of Clinical Genetics, University of Newcastle upon Tyne and Director, Northern Genetics Service

Ms Ruth Evans (until December 2002)
Formerly Director of the National Consumer Council

Dr Hilary Harris
General Practitioner, Manchester

Professor John Harris
Sir David Alliance Professor of Bioethics, University of Manchester

Mr Alastair Kent (from January 2003)
Director, Genetics Interest Group

Ms Hilary Newiss
Solicitor

Mrs Christine Patch (from January 2003)
Genetic Counsellor

Reverend John Polkinghorne (until December 2002)
Canon Theologian of Liverpool and formerly President of Queens' College Cambridge

Professor Martin Richards
Professor of Family Research, Centre for Family Research, University of Cambridge

Dr Gill Samuels (until December 2002)
Director of Science Policy (Europe), Pfizer

Mr Peter Sayers (from January 2003)
Chair of the Telecommunications Advisory Panel

Sir John Sulston
Former Director of the Sanger Center, part of the Wellcome Trust Genome Campus, Cambridge

Professor Veronica van Heyningen
Head of Cell Genetics Section, MRC Human Genetics Unit, Edinburgh

Mr Geoff Watts
Journalist and presenter of BBC Radio 4's Leading Edge

Mr Philip Webb
Member of the Board of Trustees of Genetic Interest Group

Ex Officio Member
Ms Suzi Leather
Chair of Human Fertilisation and Embryology Authority

Representatives of the Chief Medical Officers
Each of the four UK Chief Medical Officers will be able to participate in HGC or nominate a representative with observer status.

Dr Stephen Singleton (England)
Medical Director, Northumberland and Tyne & Wear Health Authority

Professor Peter Harper (Wales)
Professor and consultant in medical genetics, University of Wales

Professor Patrick Morrison (Northern Ireland)
Consultant clinical geneticist, Belfast City Hospital

Dr Rosalind Skinner (Scotland)
Principal Medical Officer of Public Health Medical Division, SEHD

Co-opted Members
Mr Robert Bestow (until December 2002) (Co-opted Member, Genetic Services Sub-group)
Director, NF (Neurofibromatosis) Association

Mr Harry Cayton (until December 2002) (Co-opted Member, Public Involvement Sub-group)
Chief Executive, Alzheimer's Society

Dr Heather Draper (Co-opted Member, Working Group on Reproductive Choice)
Senior Lecturer, Centre for Biomedical Ethics, University of Birmingham

Dr Frances Flinter (Co-opted Member, Genetic Services Sub-group and Working Group on Reproductive Choice)
Clinical Director and Consultant Clinical Geneticist, Genetics Centre. Guy's and St Thomas' Hospital Trust

Mrs Lesley Greene (until December 2002) (Co-opted Member, Horizon-Scanning Sub-group)
Support Services Director, CLIMB (formerly the Research Trust for Metabolic Diseases in Children)

Mr John James (until December 2002) (Co-opted Member, Horizon-Scanning Sub-group)
Chief Executive, Kensington, Chelsea & Westminster (KCW) Health Authority

Dr Keith Palmer (until December 2002) (Co-opted Member, Public Involvement Sub-group)
Vice Chairman, Investment Banking, N.M. Rothschild & Sons Ltd

Confederation
Dr Nigel Spurr (until December 2002) (Co-opted Member, Horizon-Scanning Sub-group)
Director, Genetic Technologies, SmithKline Beecham Pharmaceuticals

Secretariat
Dr Mark Bale, Secretary
Dr Manny Chandra
Mr Richard Pitts (until August 2002)
Mrs Margaret Straughan
Dr Sophie Taysom (from January 2003)
Ms Emma Wilbraham

The Secretariat is provided by the Department of Health and the Office of Science and Technology officials and may be contacted at:

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Annex B: Reports from information-gathering sessions

We have found it useful to build a regular information-gathering session into our meeting structure. This gives us a chance to discuss a particular issue in more detail, inviting a number of people to talk to us about varying aspects and to consider the implications for our own work. Summaries of these sessions are given below, with full reports of the meetings available on the website (www.hgc.gov.uk).

Pharmacogenetics

On 10 September 2001, we held an information-gathering session on pharmacogenetics, the study of how genetic variation can affect an individual's response to drugs. Four presentations detailed the potential health benefits that pharmacogenetics might bring, and some of the social and ethical issues raised.

We heard:

- how different versions of the same gene can affect whether people respond well, badly or not at all to a particular drug;
- how these differences can be characterised by determining SNPs (single nucleotide polymorphisms), single 'letter' changes within the DNA sequence;
- how associations can be made between a certain pattern of SNPs and a particular drug response, though care needs to be taken not to make false associations

Potential benefits conferred by pharmacogenetics include:

- identifying both the best drug and the best dose of that drug to treat individual patients more quickly and efficiently than is being done at present;
- reducing the extent of adverse drug events (ADE) which occurs when the patient reacts badly to the given drug, ADE's being estimated to cost around £2 – 4 billion a year in the UK;
- allowing development of drugs which are effective only for well-defined and small populations. Currently, drugs which are effective in the majority of people, but produce serious side effects in a minority, may have to be withdrawn.

In order to maximise the potential of pharmacogenetics, the data which is collected during clinical trials and the post-drug licensing period may need to be changed, and regulators might increasingly request this type of information when considering the licensing of a drug. Pharmacogenetics will be introduced slowly, depending among other things on the strength of relationship between genetic variation and drug response, and the health care cost of treatment failure. All the different ways in which the body can deal with a drug might not be evident during clinical trials.

The concern was voiced that pharmacogenetics might lead to more expensive drugs for the majority of people, and there might be no drugs or only very highly priced ones available to a small minority. In addition, many ADE's are due to human input rather than genetic variation: possible reasons included misapplication of knowledge regarding drug therapy; incorrect calculation of drug dose; and getting the drug name wrong.

The discussion raised several points:

- most diseases can be treated by a number of drugs, and pharmacogenetics would assist in identifying the most suitable for a patient
- diseases having the same symptoms could be shown actually to have different causes and so be treated more effectively
- for NHS to be able to deliver personalised medicine, a fundamental shift is required in how health care was delivered, with prescribing decisions being shared much more with patients and pharmacists. Doctors and the general public would need to be much more aware how genetic variation might affect drug response.
- Knowledge of the drugs a person is prescribed, together with lifestyle choices, might indicate underlying genetic traits; discrimination arising from the availability of this information must be prevented. Also, it must be remembered that genes known now to affect drug response might later be linked to susceptibility to disease.
- The genetic component contributing to ADE's should not be over- or under-estimated, and low-tech solutions and electronic prescribing systems can help prevent human error.

UK Biobank

In November 2002, the House of Commons Science and Technology Committee asked HGC to submit a memorandum on UK Biobank, focusing on aspects of openness, confidentiality and governance. We held an information-gathering session on the topic to assist in the drafting the memorandum. We heard from five speakers, two of whom were from organisations funding Biobank, on the scientific rationale underlying the project, the governance structures, issues around consent and data security, and considerations of commercial access to the genetic database.

We heard about:

- the history behind the project;
- its aim of providing scientists with a resource to allow them to investigate separately and in combination genetic and environmental factors on the risk of developing common disorders, such as cancers, and heart and metabolic diseases;
- the advantages and benefits of having such a resource in the UK, and why it has been structured as a prospective cohort study;
- how 500,000 people aged 45-69 will be able to donate a 50ml blood sample to Biobank via their family doctor;

Issues of oversight and governance

These are of fundamental importance to Biobank, and public consultation has been carried out in parallel to the scientific development of the project. This gauged public knowledge of genetics, and attitudes to genetic information, access to data, ownership, confidentiality and feedback. The management structure, consisting of a central Hub (including a scientific management committee), an oversight committee, and a number of spokes, was outlined. The ethical and governance framework which would underlie the project was still being developed, in conjunction with public consultation, and over the next six months or so, people and organisations would be appointed to appropriate positions within the structure.

Issues of consent and feedback

Concerns about large-scale databases included confidentiality, potential unfair discrimination, and commercialisation/patenting leading to loss of access. There is the question of whether broad or narrow consent should be requested; for the former to be appropriate, there must be a clear description of potential research, the procedures used, and how the project will be governed. It might be that specific consent would always be required for particular research eg establishing a cell line. Participants must be allowed to withdraw from the study at any time, and their identifiable data destroyed. Police and the courts should not have access to the database, and companies should have non-exclusive access only, possibly accompanied by some form of public benefit sharing. General feedback should be given using websites or newsletters.

Data security

Inference control/security mechanisms were explained. These operate to prevent personal data from being inferred from statistical queries, eg by monitoring the queries being asked, and ensuring that no less than 6 records are returned for any one query. Data can also be masked, eg by using relative rather than absolute values, and averaging the characteristics of a general group. Other protections would be to allow only vetted statisticians direct access to the database, informing participants that absolute privacy was not possible, though safeguards would be taken as far as possible, and using pseudonyms rather than real names. Particular problems arise with the recording of rare genetic disorders, precisely because of their infrequency in the population. To date, there have been no incidents of deliberate violations of privacy, more specific awareness in a particular situation. Any access of NHS records must be done with the person's full consent. Technical workarounds can offer some solutions but will not deal with the whole problem.

Commercial perspective

The commercial view is that Biobank's value would be greatly increased if samples were stored such that the proteins present in serum could be analysed. Because protein levels in a person change whereas their genes do not, the former could provide a more precise indication of what might be happening during progression of a disease. Proteins from case and control samples can be compared, and the ones present in one but not the other might indicate genes which are associated with a particular condition. Commercial companies expect a return on their investment, and while non-exclusive access is a very good place to begin from, without the protection being given to any intellectual property that might be generated, there will be no incentive to invest in the first place.

Matters raised during discussion include:

- Mentioning in the information leaflet given to potential participants that long-term benefits will arise in the form of better treatments and diagnostics, but that participants cannot share in any direct commercial gain.
- The need for rigorous ethical review. Research Ethics Committee (REC) approval is required for all research projects using human subjects, but there are issues of competency. A multi-centre REC (MREC) must be nominated to take overall responsibility.
- The implications of the Data Protection Act on the person's right for individual feedback would need to be clarified.
- The possibility of police access is not unique to Biobank, but to all collections of samples taken for clinical purposes. A court could issue an injunction to allow police to have access, either for a specific sample or to do a general trawl. HGC have recommended that this situation is changed, and that people are informed of the remote risk that the police might gain access to their data.

- Though there has never been a widespread public consultation on the topic of Biobank, there has been a generally positive response from the two focus groups that have been held, though it was not clear how well the ideas behind Biobank are understood. The Spokes and regional centres will be encouraged to highlight the issues once details have been finalised.

Work Plan and Work on Genetics and Reproduction

In February 2003, HGC held an informal meeting on two important topics, its work plan and genetics and reproduction. With regard to their work plan, Members discussed how they would cover issues around genetic discrimination and NHS genetic services, including the Government's Paper on Genetics; the importance of liaising with organisations such as the Human Fertilisation and Embryology Authority (HFEA), the Genetics and Insurance Committee (GAIC) and UK Biobank; and the need to engage with the public, to both provide information and facilitate discussion. The issue of education in genetics for both public and heath care professionals pervaded all aspects of the work plan.

With regard to the work on genetics and reproduction, Members heard two presentations. One was from Suzi Leather, HFEA Chair as well as HGC Member, who highlighted new developments for alleviation of infertility, and how these might impact on HFEA's considerations. The other was from Professor Neva Haites, of the National Screening Committee, who spoke about couple screening programmes in relation to cystic fibrosis (CF).

Ms Leather explained that changing public attitudes, the provisions of the Human Rights Act, and an increase in the number of conditions that can be tested for using preimplantation genetic diagnosis (PGD), meant that use of the technology is changing. HFEA would be publishing a new Code of Practice, which reflected the recommendations of the HFEA/HGC Joint Working Party on PGD (JWP), and also the fact that the law explicitly banned alternation of the genetic structure of cell or embryo, so disallowing genetic enhancement. Because of welfare of the child considerations, only therapeutic uses of PGD can be licensed, and only were benefit is conferred to the child being conceived, not to other family members. The Government have requested HFEA to consider review attitudes to sex selection and related technologies, and a report, incorporating the results of the accompanying public consultation, is expected in April/May.

The discussion that followed Ms Leather's presentation covered issues such as the need to investigate whether or not IVF (in-vitro fertilisation) treatment adversely affected children so conceived, and the difficulty in monitoring the health of a conceived child because of the confidentiality provisions of the Human Fertilisation and Embryology Act. There were concerns raised by the use of tissue typing to select embryos, even when checking for the presence of a serious genetic disorder at the same time.

Professor Haites gave details about cystic fibrosis, which is caused by mutations in the CF gene. The disease is an autosomal recessive condition, with parents in general being unaffected carriers. Couples can be offered screening by their family doctor, but screening models focus on the antenatal clinic. In the couple screening model, developed in Edinburgh, both partners are asked to give mouthwash samples after they have received an information pack and have been counselled. The woman's mouthwash DNA sample is tested first and if she is found to be a carrier the partner's sample is then tested. If both partners are carriers, they are invited to discuss options with the fetal medicine team. NSC is considering whether such a programme should go nationwide. Other priorities include ensuring that a high quality screening process is established for Down's Syndrome and the haemoglobinopathies in the first instance.

The discussion that followed noted the difficulty that JWP had had in agreeing what constituted a serious disorder, as what appeared serious to one family might not appear quite so serious to another; however use of a certain description of a disorder would affect perception by, and of, affected individuals.

Communicating Genetics

As the Annual meeting of the European Society of Human Genetics (ESHG) was held just before and at the same venue as the HGC May 2003 plenary meeting, HGC took the opportunity to hear about communicating genetics in the European context. Thus, presentations were given by Professor Gertjan van Ommen, ESHG President and President of Human Genetics at University of Leiden, the Netherlands, Dr Francesca Pasinelli, Chief Executive of the Italian genetics charity Telethon, and Dr Alison Stewart, Chief Knowledge Officer at the UK's Cambridge Genetics Knowledge Park.

Netherlands

- Much media attention has been given to human genetics, especially when the first draft of the human genome sequence was published. It is important to use meaningful images when communicating genetics; using pictures of electrical wiring in an old home or a mobile from which a number of objects are hanging can be better at conveying the complexity of genetics than showing the picture of a DNA gel. It is also important to convey accurately the risk associated with a specific gene sequence, and to have articles in popular magazines, which are far more approachable than scientific journals.
- The Internet is becoming an increasingly significant way of disseminating information about genetics, and it is important to ensure that as high a quality of information as possible is available. Doctors must be brought up to speed so that they can deal adequately with patient's queries. More than 50% of Dutch doctors have registered onto a medical information website, and there is also a popular website for schoolchildren.

Italy

- Science is not well regarded in Italy, with the lowest ratio of research investigators per head of general population in Europe. One of only two medical research charities in Italy, Telethon has two major stakeholders, the public and patient organisations. Telethon's aims are to advance genetic research and to involve the Italian public in the fight against genetic disease, by both recruiting volunteers and expanding knowledge.
- Money is raised principally through a 36-hour television marathon every year, with an audience of around 20 million viewers. The programme comprises 74% entertainment, 6% ongoing work in Telethon institutes, and 20% scientific information, with the broadcasting company having complete editorial control. In 2002, 600,000 donors raised around 23.4 million.
- Scientific information and findings resulting from funded research is sent to all donors, popular magazines and to major companies and banks, and a recent survey has shown that awareness of Telethon is relatively high. Telethon prepares briefing for the media on topics such as gene therapy and stem cell research, and has also produced material for GPs. Other projects include the Telethon Young project, which has involved 12,000 participants since 1999, and Telefona Scienza which offers a freephone service during the TV marathon.

UK

- The NHS workforce is not well prepared for the changes in the clinical services being wrought by genetics, and many health professionals do not recognise the need to improve their knowledge in the area. Some regional genetic centres and other organisations are providing training days for medical and health care professionals, and a project is ongoing to develop a national strategy for educating professionals in genetics, with a consultation document being issued next year.
- The emphasis on getting genetics on the agenda needs to come from Government, and the relevance of genetics to particular specialities underlined. However, each professional group must decide and own the process by which genetics is integrated into its area, and sufficient long-term resources should be available to facilitate this.
- The Cambridge Knowledge Park (CGKP) is focusing on the education of health professionals, with 4 regional training days being held initially, and with the aim of building up the number of people in policy-making positions who are aware of the issues. Other Knowledge Parks are offering similar resources. It is very important for NHS managers to consider the strategic impact of genetics, so they can facilitate its integration into clinical practice.

Discussion

- There is much going on in terms of genetic education, but more coherence is needed. There are some very good Internet resources for school teachers, but input is required from those directly involved in education to determine what is needed. Studies have shown that while young people are familiar with words such as gene and chromosome, they may not be totally correct in their understanding.
- It is important to teach developmental biology as well as classical genetics when explaining the role that genes play in the functioning of the organism, and to engender interest by highlighting the similarity of some genes in fruit flies, humans and other organisms.
- Communication is not so much a question of legitimising what is going to happen anyway as trying to give a realistic idea of the scope of likely developments. The public appears to favour a cautious rate of scientific development, and scientists should be clearer about what is possible and how soon it might be realised.

Annex C: Meeting of the HGC Consultative Panel – July 2002

Report back from the round table discussions on genetic testing services sold direct to the public

There was a sense that we should be setting standards in the UK irrespective of what might happen in other countries and that there should be a strengthening of the voluntary code of practice at the very least. Some suggested a possible need for legislative change in some circumstances but drawing lines between what does and does not need to be regulated is difficult. The was some support for the idea that people should be able to make their own decisions but it was also important to remember that this does not reflect the reality of the effects genetic testing can have on other members of a family. Perhaps there should be more discussion about the collective responsibilities that arise in this area of testing.

The following gives more detailed comments from the tables:

- One table had found it easier to see the problems than the solutions but the general feeling was that this was not something you could leave to the market – it needs regulation of some kind. After discussion the table reached the general position that in principle it might be nice to have voluntary self-regulation but there was a profound suspicion about the extent to which you could rely on that.
- Questions arose over who was actually going to do the regulating and who was going to check that the code of conduct was being looked at. There were also concerns that if you introduce a two tier system with some tests left to voluntary regulation and some by a legislative framework how do you make a dividing line between the two.
- Another table reached a general consensus that something should be done – they were not clear about what but felt that just letting it ride wasn't good enough. There was a general feeling that there is really no point in testing unless you can do something about it and that with this type of testing you have a responsibility to family and others as well as yourself.
- The issue of insurance was raised – this sort of testing would be mean you could have knowledge which you might not want your insurance company to have but that in some cases your insurance company ought to have. This shows that perhaps testing should be under some sort of framework. There was also a need to accept that no matter what we set up in this country people will have access to uncontrolled testing.
- One group had considered internet testing and related consent and confidentiality issues and generally agreed that education was part of a control mechanism as it would encourage people using internet services to use suppliers with quality tests and counselling etc. There were concerns about increasing GP workload – if a lot of people had tests then more will go to their GP afterwards, and what about educating GPs. And one of the group had had a genetic test on the NHS within the last couple of weeks but had had no counselling whatsoever, this highlights the importance of counselling within the NHS too.
- Another table discussed why you should want to be tested over the counter and the general feeling was that quite often it was to avoid having a record that your insurance company can have access to but there was no agreement as to whether this was a good or bad idea. Most of the table opted for option three, which is stop some tests for serious conditions being offered direct to the public and have a voluntary code of practice for the rest. Raised the point that legislation can be both a blunt and inflexible weapon to use in the area. The important thing is the need to make sure that people have the right understanding about the test they are asking for and the right understanding of the information they get.

- A further table made up of people with a range of varying experiences found that this resulted in a range of views from the libertarian through to the prescriptive in terms of allowing testing but interestingly this didn't stop a general consensus view emerging. The majority preferred option two, revising the voluntary code of practice for all genetic tests.
- One table said they were surprised to have heard earlier in the day that you can get a test for Huntingdon's on the internet. Table felt that even if you can't control what is done in other countries/over the net it is nonetheless important to set standards in this country. One of the group felt very strongly that people have a right to have tests for themselves. The other side to this is that as a lot of genetic conditions are hereditary someone exercising their rights to find out genetic information about themselves can take away the rights of other family members. Was interest in the fact that HIV is testing is banned and why differentiate between HIV and a genetic condition.
- Another group commented that this country has moved towards a greater emphasis on individual rights but other societies would take a family orientated view and many of the issues in genetic testing relate to whole families and not to individuals. Also that a genetic disorder can have its greatest impact on the healthy members of the family so a parent or son or brother might feel their lives have been impaired where the person directly affected may not. This would be important in deciding where to draw lines on what is or is not a significant disorder. The question of quality control was also raised – people will assume that the test is correctly carried out but the less regulated the area is the more likely you are to get wrong results so need to guard against this.

Report back from the round table discussions on genetics and reporduction

A wide range of views were expressed, which it is not possible or useful to try and summarise. The following is taken from the report back from HGC Members at the meeting and is intended to give a flavour of the discussions and the points raised.

- Several reported there was not a great deal of unanimity round their tables. This was highlighted for example, by two people who mentioned their experiences of giving birth to children with genetic disorders and one would go through the same again and the other would not. Some commented on the wide range of opinions expressed and that this was based on a wide range of experience and people came to their view based primarily on their own experience. And one table reported a perplexed and perplexing discussion, which did not reach many conclusions as these are such hard problems.
- A number of tables stressed the importance of personal choice, for example one table agreed that testing should be a matter of personal choice but this should be informed choice. This raised the question of education and the group felt there should be education in such matters from school age so that people have a chance to think about these sorts of issues in case they need to make such a decision in the future. Cultural and religious views were felt to be very relevant in the decision making process and that different people in different circumstances will have different interpretations of what is a serious disorder. They felt the term designer baby should be dropped because there is no such thing and it is not something they saw as desirable.
- One table did not have a great deal of sympathy for the view that preventing the birth of children with genetic disorders was an implied slur on those who are born with disorders and felt the emphasis should be on personal choice. They also hoped that more education would lead to less discrimination against people who have a genetic disorder and therefore some of the fear in this respect would disappear. Hopes that research will get beyond preventing the births of children with genetic disorders and provide cures were seen as possible way round this issue, but that this was a long way into the future.

- One table said that the area of personal choice should be kept wide and discussed ways of defining a disorder as serious and felt that criteria such as will the child ever become independent could be useful but realised that not everybody will agree with this. Talk about things on a case by case basis may make it easier for people to reach an agreement than when discussing generalities.

- Another table felt that there was a wide spectrum of conditions involved – with untreatable conditions resulting in early death at one end and much less serious conditions such as cleft palate at the other – and all the difficulties lie in the middle of the spectrum. The problem was how to work out where one changes into the other, particularly as in different contexts and different circumstances the lines are rightly drawn in different places. Table also discussed who the interested parties were in this. Parents are an interested party of great importance but are not the sole party, and the foetus is also important but is not able to express an opinion or be represented directly in all the decisions. Also the medical profession need to feel that they are asked to do things that are ethical and justifiable and there are the views of society. People can face these very difficult decisions out of the blue and have to make quick and stressful decisions, so there is a role for society to have a say and provide an ethical balance. If society does have a say then it also has to accept the responsibility that stems from that, which comes back to issues of resources and provision of services etc.

- One group, while recognising the very broad range of views expressed which reflected the wide range of experiences around the table, generally felt that prenatal diagnosis was useful, not necessarily in reaching choices, but in preparing people for an eventuality. Much of the discussion centred on being given the choice of information to prepare people to continue with pregnancies and build for the future. They also discussed the fact that there are complex hurdles involved in PGD and it is not something taken lightly and expressed some difficulty in getting to grips with the concept of designer babies – what does this really mean?

- Another table felt that whilst they understood that parents with disabled children may feel that they can't bear to have other children similarly affected, it was important to look at why they did not want those children. They saw this as being because the emphasis is on the impairment and not on the social impacts, and it is difficult bring up a disabled child in today's world because of a lack of services. They felt that reproductive choices focussed on the impairment rather than on the person and that there was too much assumption and prejudgement about another's quality of life. The table felt behaviours would have to be changed through structures rather than hoping for a change in attitudes, which would take too long.

- One group discussed the prospect for therapy, which would have an impact on these decisions. It was suggested that if you take comments round the table from people with these sorts of experience then will always find someone to counter someone else's opinion. The table saw a need for a greater capacity, perhaps through the HGC's website, for a chat room type interaction between members of the Panel. This would mean a greater discussion of views and might generate a distillation of views from several members.

Annex D: Published responses and memoranda

HGC response to "*Human Bodies, Human Choices*" consultation report on the law on human organs and tissues in England and Wales

1. The Human Genetics Commission has given attention to *Human Bodies, Human Choices* and would like to make a number of observations. The Commission believes that this review has been particularly timely and we hope that the work that we have recently undertaken on personal genetic information – which is acknowledged in the consultation report – will be of assistance to the Department of Health and the Welsh Assembly Government in its task of recommending legal reform in this area.

2. The Commission has recently made a number of recommendations to Ministers relating to personal genetic information which could possibly be acted upon in the context of this broader review of the law relating to human tissue. Indeed, the Commission believes that it would be a lost opportunity if legal reform relating to human tissue failed to address very specific issues surrounding the handling and use of human DNA.

3. We address below a number of points raised in the consultation report. Our response is arranged following the order of the sections in that report.

Section 3 – Present legal framework

4. The consultation report points out that the legislation on human tissue is ripe for review. As far as human genetic material (DNA) is concerned there is little specific legislation. There is a strong case for ensuring that future human tissue legislation includes at least some recognition of the particular problems associated with human genetic material. Most human tissue contains genetic material and interest in the uses of genetic information are increasing with advance in genetic knowledge.

5. **Question 3A.** We draw your attention to the current comprehensive consideration of human genetic information by the Australian Law Reform Commission and the National Health and Medical Research Council (available at www.alrc.gov.au)

6. **Question 3B.** We believe that it is important to acknowledge the cultural significance not only of the tissue itself but also of the information that the tissue contains. Any regime constructed to deal with human tissue should therefore take account of sensitivities associated with personal genetic information. In this respect we refer to the discussion of this issue in our report Inside Information (paragraphs 1.17-1.20). We concluded that

   The fact that genetic information is considered by many to belong to a particularly sensitive and private category of information may merit giving it enhanced status [in order to] address the sense of possession which many feel about their genetic information [and] the sense of violation of privacy if personal genetic information is wrongfully used.

Section 4 – Scope of the review

7. We note the contents of the draft Code of Practice on the Import and Export of Human Body Parts. For convenience we include here our comments on that document.

8. The import and export of human genetic information is an important feature of diagnosis and research into human genetic conditions. We consider that any Code of Practice should not inhibit this activity inappropriately. However, we question the exclusion of DNA from the scope of this Code (paragraph 4). The obtaining of human genetic material from overseas has raised certain ethical issues and is in some countries a matter of controversy. We think that there should be

some ethical oversight of how imported human genetic material was obtained in the country of origin. **We would recommend that consideration be given to this issue before the Code is finalised.**

Section 5 – defining organs and tissue

9. Although we believe that future human tissue legislation should deal with certain specific genetic issues, we appreciate that some of the rules relating to human tissue itself will not be appropriate for human genetic material. In particular legislation must avoid placing inappropriate restrictions on laboratory work on human DNA. Such DNA may have been purified by recombinant DNA techniques and is replicated in cell culture. It does not contain recognisable human tissue.

10. For this reason, **Question 5A** is very important. The use of the term 'human materials' would cover human DNA even if it were isolated from human cells, for example using recombinant DNA techniques. It is therefore probably too broad.

11. **Question 5B.** We agree that the scope of the legislation must be carefully defined and that certain forms of human tissue need not be treated as being of great significance (the examples given in the report are of nail clippings and hair cuttings). However, we are concerned about some potential uses of discarded bodily materials of this nature. In our report 'Inside Information' we pointed out how discarded bodily materials could be subjected to genetic analysis for improper reasons.

12. We note that Section 4 stresses that the new legal framework should provide a statutory basis for regulating all aspects of obtaining, storage, use and disposal of all tissue and organs. We believe that genetic analysis is a very significant potential use of human tissue and must be specifically addressed. If the proposed new legislation excluded such materials from its scope then we believe it would be important to make separate provision relating to their subjection to genetic analysis.

13. One possibility is that the new law could draw a distinction between **removed tissue** (such as clinical samples or research samples) and **discarded tissue**. Removed tissue would be subject to appropriate regulation to ensure appropriate safeguards on consent, confidentiality and respectful disposal as set in later sections of the consultation (see below). Discarded tissue, however, would largely fall outside of the scope of the new Human Tissue Act unless it was to be subject to genetic analysis that was not for a lawful purpose (such as by the police). We believe that it is important to strictly control the unauthorised genetic analysis of removed and discarded tissue. Irrespective of whether there is any wider disclosure of genetic information, the principle of genetic privacy that we have adopted would mean that the non-consensual analysis of personal genetic information would constitute an unwarranted intrusion of privacy.

Section 7 – Who would give consent

14. We note that on page 41 concern is expressed about one of the recommendations made by the HGC in Inside Information (p85). This recommendation was that in the absence of any consent given in lifetime to the *post mortem* genetic testing of tissue it would acceptable for such testing to take place on basis that the dead person would have consented had consent been sought during his or her life. This presumption would only be made in those cases where such testing was necessary for the clinical care of a living relative. We believe that this is compatible with the consent principle which the consultation report advocates in Section 6. The general principle stated there is appropriate for research use of tissue which does not directly benefit a living relative. In such circumstances it is reasonable to insist on obtaining consent from the next of kin.

15. Different considerations apply where a living relative has a strong interest in the testing being carried out and we believe that this interest may be recognised without diminishing our respect for the consent principle in general.

16. The question of property rights of a body or its parts is a difficult one that may have benefited from a more detailed treatment in the consultation report. However, whilst we have considered this in some detail in our work on personal genetic information, it has proved difficult to establish any clear recommendations. In our report on personal genetic information we concluded that best practice in research required that factors such as the fate of samples and commercial involvement should be made clear when seeking consent. In particular, whilst we recognised the value of the "gift" interpretation of samples we also noted the importance of considering any implications for intellectual property and commercial access to research and samples.

17. The later sections of the consultation report refer to property rights and cell lines or tissue engineered products (section 17). In our Inside Information report we endorsed the approach taken by the House of Lords Stem Cell Committee that no specific property rights or consent should apply to cell lines developed from a sample. However, this was on the proviso that the position was made clear before seeking consent and that individuals were free to decline to donate samples.

18. We also note the related point and question **17G** about any system that might be put in place to ensure that the donors receive some benefit. We have considered this in some depth in relation to genetic research databases. We have recognised the importance of commercial involvement in such research and the public unease about commercial profit from medical research. We concluded that in return for altruistic donation to research programmes there should be some benefit to the participants, or the wider community from which they are drawn. However, we felt that such benefit-sharing mechanisms could best be established in the light of the particular circumstances. We therefore recommended that national benefit should be taken into account in determining the terms on which commercial involvement was granted to research databases or human tissue samples.

19. Therefore, in response to questions **7P** and also to **17F/G**, we would agree that the legislation on human organs and tissues provides an opportunity to clarify the issue of property rights for human tissue. We believe that any framework should ensure that those consenting to the use of human organs and tissue are provided with clear information about the eventual use of their samples and to whom the benefits accrue.

Section 8 – Defining consent

20. We have conducted a detailed examination of the legal basis of consent in the context of genetic information. We have adopted the principle of consent as one of the secondary principles underlying our central principle of respect for persons. Therefore, whilst we note the possible alternatives to the use of term in any new legislation, we believe that "consent" has gained a wide acceptance in UK and international law and should be retained.

21. However, we note the concerns expressed about the giving of detailed information to relatives in requesting consent for a post-mortem examination. There are similar concerns in the area of pre-symptomatic genetic testing. In our report we examine the concept of a "right not to know". We conclude that whilst people may not wish to be burdened with bleak knowledge, there are equally circumstances in which such information is necessary for them to exercise personal autonomy. We therefore prefer the term an "entitlement not to know" in recognition of the fact that there may be circumstances where this must be over-ridden. We would hope that it would be possible to adopt this approach, perhaps together with the concept of "authorisation" that is recommended by the review in Scotland, in the legislation, Codes and guidance that are being prepared.

Section 9 – What is removed and why

22. This section details a number of areas in which human tissue may be retained or used (paragraph 9.4). In response to **Question 9J** about non-clinical or non-scientific uses of human tissues, we wish to comment about the possible omission of genetic testing services that are supplied direct to the public (including genetic testing services, DNA paternity testing or genealogical analysis). Most of such services require the obtaining of a sample of buccal cells, or more rarely, blood samples.

23. We are in the process of consulting on the regulation of such services, but at present we only note that they are regulated by two voluntary Codes of Practice published by UK Health Departments. There is also a variety of general consumer protection and data protection legislation that would apply to such services. We have set out further details in a consultation paper (enclosed for information) and we will comment elsewhere about our emerging conclusions.

24. We would also point out that there are a significant number of commercial companies who collect tissue samples directly or via NHS clinical centres for use in research. We have no reason to believe that these are not collected ethically, but they would represent another commercial sector that might be affected by any new controls. We also believe that these activities raise very important and potentially controversial issues of property and ownership of human tissues, which we comment on below.

Section 10 – Training, education and research

25. We have noted the draft interim statement of principles for clinical research (paragraph 10.13) includes the requirement that all research using human organs or tissue must be approved by a properly constituted research ethics committee. We would strongly support this and in our report on personal genetic information we similarly concluded that best practice should require that all genetic research on human non-anonymised tissue samples or bodily materials should be subject to review by an independent research ethics committee and should be monitored for compliance through clearly specified arrangements. We have also recommended that the Government should encourage relevant research institutions, professional bodies and funding organisations to establish clear policies aimed at ensuring compliance with the emerging best practice in ethical research.

26. In response to **Question 10B** we suggest that there are certain categories of genetic material that might alter their legal status in a research context. We have concluded that genetic research on anonymised human samples, especially established cell lines or information derived from anonymised samples or medical records, should not automatically be subject to subsequent ethical oversight. In general we suggest that any sample of human material that has been anonymised should be excluded from rigorous oversight, provided that the sample was collected in an ethical manner. However, we believe that in doing so there should be adequate safeguards to ensure that such samples cannot subsequently be linked to a living individual.

27. We have also noted the comments here about HGC's report in relation to genetics research and look forward to the outcome of the review.

Section 11 – Oversight and compliance

28. We have commented above that the scope of the Human Bodies, Human Choices review appears to include commercial genetic testing companies and DNA paternity testing. The nature and scale of such activities may be pertinent to the possible role of a single oversight body and the requirement for licensing versus registration (**Question 11A**).

29. We are still awaiting all the responses to our consultation and therefore we cannot supply any precise figures for the scale of such activities to supplement those in paragraph 11.11. However, in our previous report we have noted that approximately 10,000 paternity tests are commissioned per year, many of these related to the work of the Child Support Agency or the Home Office Immigration Directorate. The majority of tests are conducted by 3 or 4 commercial laboratories.

30. We have paid considerable attention to the issues raised here in relation to our current review of the supply of genetic testing services direct to the public. We have not completed our review so we therefore offer the following comments as an interim response to **Question 11D**.

31. In our consultation we have identified four possible options:

- Do nothing – and let existing consumer protection legislation that covers other forms of home testing cover genetic tests too.
- Revise the voluntary code of practice for all genetic tests – this would advise on the best way to offer the tests, what information people need before they take them and how the results should be stored and protected.
- Stop some genetic tests being offered directly to the public and set up a voluntary code of practice for the rest – this would put restrictions on some types of test (most likely those for serious and life threatening disorders) and mean people have to talk to a health professional before they have these tests.
- Stop all genetic tests being offered directly to the public – this would make it an offence for anyone who was not a health professional to offer a test directly to the public.

32. We are increasingly attracted to some form of hybrid system that includes an element of statutory regulation of the provision of genetic testing services to the public. This might be at the level of the new Human Tissue Act that imposes requirements for consent, storage and disposal of tissue samples. There could be a requirement for a licence or registration and compliance with the relevant statutory Code of Practice. Such Codes might be based on the two existing Codes of Practice (the Advisory Committee on Genetic Testing (ACGT) 1997 "Code of Practice and Guidance on Human Genetic Testing Services supplied direct to the public" and the "Code of Practice and Guidance on Genetic Paternity Testing Services" issued by UK Health Departments in 2001. They include specific requirements relating to laboratory standards, advertising and promotion, pre- and post-test counselling as well as record keeping.

33. We have noted that the existing Codes are not supported by effective compliance-checking and enforcement mechanisms. This would be an important aspect of any future system and we have been impressed by the work of the Office of Fair Trading that is soon to be responsible for approving voluntary Codes of Practice.

34. We are, however, aware that the Lord Chancellor's Department is responsible for regulations that require accreditation of laboratories that wish to be approved for court-directed paternity testing. The accreditation process requires compliance with the otherwise voluntary Code of Practice.

35. We stress that these are our initial views based on our deliberations to date and taking into account the proposals in the Human Bodies, Human Choices document. We are aware that there are several other strands to our work, not least the mechanism for evaluating the quality and utility of genetic tests. However, the HGC would be extremely interested in further discussions about future legislation on these matters.

Section 12 – Penalties for non-compliance

36. We have not considered in detail all of the issues about penalties and who commits an offence in this section. However, we can offer the following comments about the gradation of penalties in **Questions 12E-F**. In the light of publicity around HGC's earlier recommendation for a new offence of the non-consensual testing of DNA, we have discussed the concerns of men who wish to conduct a paternity test on children. Their ability to give a valid consent on behalf of the child will depend on whether they have parental responsibility for the child. The current law is to be amended by the Adoption and Children Bill, which is currently before Parliament. However, we believe that any new offence should take into the possibility that there should be graduated penalties in any new offence, such that testing for personal reasons without a valid consent (e.g. testing of a child by a putative father) might attract a fine. On the other hand, testing by third parties who have no personal connection to the person being tested (such as the example of testing of dental floss by a private investigator) might be liable for imprisonment or a higher fine or both.

Section 15 – Fetal tissue

37. We note the comments in Section 15 about the need to review the Polkinghorne guidelines. The Commission would be very interested in commenting on aspects of such a review that may impact on the use of fetal tissue in genetic research. At this stage we do not have any comments to make on the questions here. However, we would note that in the response to our 'Whose Hands on Your Genes' consultation the Royal College of Obstetricians and Gynaecologists states that fresh tissue rather than stored, is vital for a great deal of research. Although this is acknowledged by the DoH guidelines, the Royal College maintain that this 'principle of separation is more difficult to maintain at local level and creates barriers to research'.

Section 17 – Cell lines and stem cells

38. We would broadly support the requirement for ethical obtaining and establishment of cell lines as set out in paragraph 17.7. In response to **Question 17A-B** we draw attention to the remarks in our report on personal genetic information. We concluded that

    Genetic research on anonymised human samples, especially established cell lines or information derived from anonymised samples or medical records, should not automatically be subject to subsequent ethical oversight. We recognise that human genetic research cannot reasonably be categorised into those projects which may cause concern, harm or distress to the research and those in which the subject has no particular interest. There is a clearly a spectrum of concern and there will be grey areas upon which researchers may wish to seek advice from an established independent research ethics committee, such as an NHS REC.

39. In response to **Question 17C** we have also highlighted the future potential for anonymised cell lines to be associated with an identifiable person by via DNA fingerprinting or de-encryption of computer and manual records. This is one of the scenarios that led us to recommend the new offence of non-consensual testing or analysis of DNA.

40. The above issue may arise in connection with stem cell lines. We have noted the important debate surrounding the derivation of embryonic stem cells. We also note that the Government does not consider that the use of isolated ES cell lines requires additional ethical oversight. We have briefly considered the wider use of stem cell lines. We note the comments in Section 15 about the need to review the Polkinghorne guidelines. Many of the issues that relate to the derivation of and subsequent use of fetal stem cell lines may equally apply to the use of embryonic or adult stem cell lines. In particular, there is the possibility that the biological 'parents' of the foetus or embryo may be identified by subsequent research or DNA analysis. This may, indeed, be a requirement of medicines regulations designed to ensure that the

source of biological medicines remains free from serious conditions such as HIV infection or CJD. We do not necessarily believe that this requires any additional oversight mechanisms, but we consider that the issues surrounding stem cell lines from any source remain under review.

41. We have noted above our views on question 17F-G about property rights and benefit sharing.

We would like to conclude by stressing the importance of this review in establishing a comprehensive legislative framework for the appropriate control of the myriad uses of human organs and tissue. We have stressed in our report the importance of proper controls to ensure that there is a balance between the interests of the individual and of the wider biomedical community and society. If there is any further help that the Commission could provide to this review we would be happy to consider it.

Human Genetics Commission
11 October 2002

HGC memorandum to the House of Commons Science and Technology Committee on the Medical Research Council/ Wellcome Trust/Department of Health UK Biobank study

1. The Human Genetics Commission welcomes the opportunity to assist the Science and Technology Committee's consideration of the setting up of the Biobank genetic research database. HGC has considered some of the issues raised by this major research programme as part of our consideration of the protection of personal genetic information. In May 2002 we published a report entitled *"Inside Information – balancing interests in the use of personal genetic data"*.

2. In preparing our report we took account of a large number of very detailed contributions from a wide range of individuals and organisations. These included the Medical Research Council (MRC) and the Wellcome Trust who were considering the funding for the Biobank study. The decision to fund the study was taken in late April 2002 when our report was in the final stages of preparation. We accepted that the Biobank Funders (i.e. representatives from the MRC, Wellcome Trust and Department of Health) would not be able to contribute to detailed consideration of some of the Biobank issues until funding was secured. Therefore, our report was necessarily rather general and was intended to provide a basis for more detailed consideration by the Funders in collaboration with HGC and others.

Summary of HGC's position

3. Before considering the detailed consideration of the proposals for UK Biobank, the Commission would like to formally record that we believe that this is an extremely important and valuable research project if the benefits of advances in genomics are to converted into a more detailed understanding of complex diseases. We believe that this is possibly a unique opportunity and that it must succeed. In particular, we consider that the UK Biobank is such a long-term project that there should be sufficient investment in its establishment to allow as-yet unasked questions to be answered 5, 10 or 20 years hence.

4. We have considered some of the difficult detailed aspects of the protocol and governance arrangements. The UK Biobank has joint funding from the MRC, Wellcome Trust and Department of Health which was approved on the basis of carefully developed *draft* protocols for scientific and technical organisation, as well as *provisional* proposals for consent and ethical oversight arrangements.

5. Following the funding decision all aspects of the protocol need to be finalised, but this can only be achieved once the complex organisation is more established. Arrangements are in progress to put in place the agreed management structures which include

   - Appointment of the Chief Executive;
   - Selection of the 'hub and spokes' centres which will then work together to develop more detailed final proposals in all these areas.

6. The development of final protocols is strongly dependent also on the appointment of:

   - the *scientific management board* (SMB) which will co-ordinate the development of the scientific and technical aspects of the final protocol
   - an *independent oversight body* (IOB) which will play a key role in setting the ethical and societal guidelines for the project.

7. We note that in some cases there are "chicken and egg" questions that cannot be addressed until the structures of the Biobank are established and staff appointed. However, we believe that it is important to consider all possible aspects, even if they cannot easily be resolved at this stage.

8. We remain extremely interested in the detailed consideration of these important elements and remain willing to consider and comment on any proposals. We also may need to reserve the right to provide advice separately to Health and Science Ministers if we feel that there are any important aspects that require more formal consideration by HGC.

9. Finally, we remain concerned about the importance of ensuring confidentiality of material and data in the UK Biobank. We believe that there should be a clear Ministerial statement to confirm that there will be no police access to the database. However, we also recognise that it will not be possible to ensure total confidentiality and therefore we believe that there is a much broader need to prevent research volunteers suffering unfair discrimination in the unlikely event that identifiable and sensitive genetic information is released from the UK Biobank.

HGC's role in setting up the UK Biobank

10. The early work on the establishment of a large population biomedical cohort began shortly before HGC was established in December 1999. The issues that were raised by the proposed study, and by plans in Iceland and other countries, was behind the decision by HGC to consider the protection of personal genetic information as its first main work item. We also collaborated with the House of Lords Science and Technology Committee in the preparation of their very thorough report on Human Genetic Databases which was published in April 2001.

11. The HGC Secretariat was involved in the initial scoping work by the Wellcome Trust and MRC. Individual members of HGC have also been involved in the preparation of the Biobank protocol and in the consideration of the practical (for example, in primary care) and ethical issues that it raised. This culminated in a major workshop on ethical issues in April 2002.

12. Since mid-2001 the Biobank Funders have held a series of informal meetings with the HGC Working Group or Business Committees to discuss areas of common interest. However, HGC has been slightly frustrated by our inability to consider some specific aspects of Biobank in public HGC meetings until such time as a formal funding decision was made. Whilst we respect the difficulty that Funding bodies may have had discussing the details during these early stages, this has hampered our detailed examination of some of the issues.

13. That aside, we have considered the available information on UK Biobank in the preparation of the HGC report "Inside Information; balancing interests in the use of personal genetic data" which was published in May 2002. A summary of the relevant general aspects of the report is at **Annex A** to this Memorandum.

14. Since the funding decision in April 2002 we have been able to have more detailed discussions with the Funders. On 19 November 2002 we held an information-gathering meeting at which representatives from the MRC and Wellcome Trust outlined progress on the scientific protocol and the governance arrangements. This memorandum reflects the points raised in that meeting and the discussion by the Commission on 20 November 2002. Details of these can be found on the HGC website (www.hgc.gov.uk)

Summary of points discussed on 19 November

15. The scientific validity of the study design for recruitment strategies is still being debated. This will need to be finalised, along with sampling techniques, sample preparation and storage, and genotyping and phenotyping procedures. This will be a task for the SMB in conjunction with the hub and spoke scientists, whose appointment is still pending. We understand that these will not be in place until mid-2003.

16. Two points in particular are worth highlighting. We heard from Dr Andrew Lyall about the importance of proteomics (the study of the proteins produced by living organisms) as a means of discovering the link between genes and environment and of providing targets for drugs and antibody therapies. There are currently no plans to take or store samples in Biobank in a way that would allow later proteomic analysis. This is partly for reasons of economy – each sample would require additional treatment to preserve the proteins. The arguments for considering proteomics are similar to those for the establishment of cell lines, namely that future researchers will benefit from a far-sighted initial investment in establishing the UK Biobank.

17. We also heard concerns from Professor Alan Wright about the possibility that the methods used to identify links between single genes and disease might not work for diseases in which a large number of genes had a minor effect. He considers that this possibility means that the Biobank should be more broad-based to allow its use to address other scientific issues, not least gene discovery. This would include, among other things, ensuring that suitably sized subgroup is included with available siblings, and where possible their parents, within Biobank. Finally, the lifestyle information gathered by Biobank could potentially be very important for wider health and social science research. In setting up the Biobank the Funders should talk to a wider range of clinicians (such as gerontologists and social geriatricians) and academic researchers who are concerned with issues in later life, including retirement, family support and kin relationships.

18. We are concerned to ensure that the Funders consult widely with other groups and companies to establish whether simple changes at the outset could dramatically improve the value of Biobank for a wide range of research. If these changes require additional funding or infrastructural changes, then the Government, biomedical charities and industry should be approached for additional funding.

19. Complex technology development is required for data storage and retrieval. There is currently no way to access NHS patient records, a key component of the UK Biobank programme. Even if this can be developed soon, the UK Biobank will need to make allowances for routine direct monitoring and audit of GP records. We were also told by an expert information technology developer, that achieving more or less complete data security may be impossible using existing encryption techniques. He suggested that the situation could be improved, though not made completely secure, through use of inference control for people accessing the databases. We cover this in more detail below.

HGC's recommendations large-scale genetic databases and UK Biobank

20. The general principles that were established in the preparation of the Inside Information report (Annex A) were important in shaping the Commission's conclusions and recommendations on various aspects of the use of personal genetic information in research. The report covered in some detail the various types of genetic research and the continuum in some cases between clinical care, research and commercialisation. The report sought to make clear that research might have direct or indirect benefits to the individual participants. Some research may only be predictive at the population level and the benefits may be distant or uncertain, however, all such 'blue skies' research is vital. Therefore the report concluded that as many people as possible should feel able to participate in genetic research, confident of the security and confidentiality and that ethical standards will be upheld.

21. Our conclusions apply to all forms of genetic research. However, there are some aspects of large research databases that were felt to require more detailed consideration. Our report was drafted in the light of initial discussions on the Biobank study, on similar databases in other countries – notably Iceland and Estonia – and also on previous UK experience, particularly of the databases considered by the House of Lords Science and Technology Committee.

22. There were felt to be three main additional considerations:

- Consent and confidentiality
- Ownership and benefits
- Oversight and compliance

Consent and confidentiality

23. Our report also considered in some detail the arrangements for seeking consent. In particular, the HGC debated the possibility of seeking blanket consent to research studies to avoid potentially intrusive approaches for fresh consent at a later stage. The report noted the practical need for broad consent in the face of a fast-moving technology. It concluded that it is acceptable to seek general consent in cases where there is to be irreversible or reversible anonymisation of data and samples. The report also concluded that best practice required that the consent should make clear the arrangements for subsequent withdrawal from a research study.

24. One central theme of our discussions on the scope of consent was that it should not deceive the participant about what might be done with their sample or information. This applies to some of the potentially controversial aspects of the UK Biobank, particularly data security and third party access and ownership or intellectual property. It was also considered important to draw attention to possibly more controversial research plans, for example the possibility of creating cell lines for more intensive biological analysis from some participants.

25. The organised collection of a large number of tissue samples and identifying information (including detailed information on lifestyle factors) as well as the ability to link information about genetic characteristics to information about past exposures or diet and to future health are crucial to the function of the UK Biobank. Such information is potentially of interest to other individuals, to commercial companies (outside of medical research) and to law enforcement bodies. We were concerned by the comments made by the Information Commissioner about the exclusions in the Data Protection Act (DPA) which may allow access to such information. There is considerable experience of large research studies, but the scale and duration of the planned Biobank may raise new considerations about ownership of the information and commercial access to samples and consequent intellectual property rights.

26. It was clear from our consultation and opinion surveys that the issue of confidentiality of the information, and access by commercial interests, was of major concern. The consultation revealed some disquiet over this matter, but the report stressed the important role of commercial sponsors. The report concluded that the question of commercial involvement needs to be clearly explained when seeking consent. In some cases it might be necessary to limit the degree of commercial involvement, for examples only to companies engaged in health-related research.

27. We have spent considerable time discussing the question of personal feedback of research results. However, there is a wider concern about the possible use of the access provisions of the DPA which may allow an individual to request access to their own data and even results of genotype analyses. Such requests may be from pure curiosity or they may be as the result of pressure from family members, employers or others. We are of the view that such disclosures should be discouraged, not least because there is a risk of impersonation being used to deceitfully gain access. We understand that the Biobank Funders have sought an opinion from leading Counsel and we would be very interested to consider the implications of that advice. We draw your attention to this potential uncertainty and the possible need for legislative clarification.

Confidentiality and data security

28. In our report we addressed the anonymisation of genetic information (i.e. interrupting the link between identifiable individuals and their genetic information). This may be an important factor in obtaining consent to a research study. We heard that in the light of "DNA fingerprinting" techniques it was never possible to truly anonymise genetic material. However, we concluded that methods were available to ensure that irreversible anonymisation was possible. We recommended that the Government funded research into the techniques of encryption to ensue data security. Ultimately, the report concluded that the procedures used to ensure anonymisation and confidentiality are carefully considered and explained to the research participants when seeking consent.

29. We were impressed by the points made by Dr Ross Anderson from Cambridge who felt that there was a risk that encryption could become a technical 'comfort blanket' that led to complacency about other aspects of data integrity. Dr Anderson commented on the importance of inference control that would prevent information being obtained by a series of overlapping data queries. The techniques are well established in other countries for handling health records and census returns. One important aspect of this is a cut-off so that any query result that gave a result on fewer than a specified number of individuals would not be allowed.

30. We feel that we should raise this matter with those responsible for establishing electronic NHS records and look for assurances that there will be effective mechanisms to prevent unauthorised disclosure. In particular, the operation of such systems needs to be properly monitored and effective action taken if there are breaches.

31. As well as planned access by outside agencies, there is also the more likely scenario of weak or ineffective procedures and systems by the various elements of the Biobank study. Our report therefore recommended that the operators of the Biobank (and other research databases) should be required to take rigorous steps to ensure that unauthorised access or disclosures are prevented. For example, there should be careful arrangements for vetting staff and for ensuring that maintenance of confidentiality should be a condition of employment. Any material breach of this should result in dismissal.

32. Because we concluded that complete confidentiality of databases is impossible to achieve, we wish to repeat our recommendation that misuse of Biobank data information should be made a punishable offence. We have recommended the creation of a criminal offence for the unauthorised or non-consensual testing or analysis of genetic material. We sincerely hope that this will be something that the Government will act upon, and we believe that this will be a further deterrent against the unauthorised disclosure of information.

33. Ultimately there will remain a remote possibility that identifiable information will be released from the UK Biobank and that this must be clearly explained when seeking consent. The safeguards to ensure confidentiality will need to be clearly spelt out, along with the possible nature and type of breaches of confidentiality.

34. However, it should be recognised that if information is released the research participant should not suffer harm in the sense of unfair discrimination by employers or insurance companies. We have noted the clear statement by the Association of British Insurers, the UK Forum for Genetics and Insurance and the British Society for Human Genetics that insurers will not consider results from research studies. We would also recommend similar assurances from some of the key employer's organisations. In the longer term we have clearly recommended to Government that there should be consideration of specific legislation to prevent unfair discrimination, and we await their response.

Access by police and law enforcement agencies

35. In our view UK Biobank will be so much larger and more organised than other research or clinical databases that it may be attractive as a source of police intelligence. It is feasible that the same genotyping methods (SNP profile) used on Biobank samples could be applied to a sample from a crime scene. The police might request (informally or via a warrant) that the SNP profile from the crime sample is checked against samples in the Biobank in order to see if there is a match. There would need to be a further step to de-encrypt the identifier to produce a name or other intelligence.

36. Our report recommended that genetic research databases established for health research should not be used for other purposes and that this should be put beyond doubt, by legislation if necessary. We are still awaiting a formal Government response to this and other recommendations. We have detected some concerns that there may be circumstances in which it would be in the best interests of society to give the police access to help to solve serious crimes. However, we must balance against this the potential impact on such an expensive and long-term research programme as the UK Biobank.

37. We continue to believe that this matter should be put beyond doubt well before volunteers are sought for UK Biobank. We accept that legislation may not be possible in a reasonable time frame and that there may be questions about how any legislation can cover all conceivable biomedical research databases. We feel that this is sufficiently important to merit a statement to Parliament by the Home Secretary or other senior Minister. This should clearly state that the police would never request access to the UK Biobank, or failing, that make clear the circumstances under which police access might be sought for particularly serious crimes. This information could then be given to individuals when seeking consent.

Ownership and benefit-sharing

38. Any large database such as Biobank must make provision for authorised access to data and samples by collaborating researchers. We have considered one aspect of this that seemed to raise concerns amongst some respondees. This is the question of access by commercial organisations, and the linked question of the ownership of the intellectual property rights that may accrue.

39. The example of the deCODE database in Iceland was raised by a number of respondees to our consultation and in the evidence presented to the House of Lords. The HGC felt that the question of who stood to benefit was complex. Biomedical research, and hence all of society, stands to benefit in the long term. However, much of the work will be done by commercial companies who stand to make profits from their investment in genetics research (which is bound to be more extensive than any one database). HGC acknowledges both the importance of commercial companies in biomedical research and also the moral concerns of individuals who make an altruistic contribution to research. It concluded that altruistic acts by individuals might be encouraged if companies using the data were prepared to reciprocate this community interest. It concluded that population genetic databases, established with and supported by public funding, contribute a national asset. Whilst acknowledging the need for commercial access and a reasonable period for commercial opportunities, the terms of access should be such that there is at least some benefit to public-domain biomedical knowledge.

40. We continued this discussion in the light of the detailed arrangements for the setting up of Biobank and of the latest position of the MRC, the Wellcome Trust and Department of Health for commercial access to the data. The concept of benefit sharing should be considered again, as this idea was attractive to several respondents surveyed for their views on donation of samples for analysis. There are obvious practical difficulties of any form of financial benefit-sharing where there is not readily identified link between access to Biobank and long-term financial returns.

41. We therefore consider that the benefit must be considered more generally as the sum of medical knowledge. We strongly support the proposed approach that commercial access should be granted on a non-exclusive basis. The experience of the publicly-funded Human Genome Project was noted and supported by HGC.

Future oversight of UK Biobank

42. All the above developments need to be in place before consent details can be addressed and together with other ethical issues the appropriate ethics committee can be approached to grant approval. The detailed ethical arrangements for research projects require consideration on a case-by-case basis. There is a recognised mechanism for considering many of these issues in the form of Research Ethics Committees (RECS). Our report recommended that the Government take steps to require all research on human non-anonymised genetic material is subject to review by an independent research ethics committee and is monitored for compliance through clearly specified arrangements. This is something that the Funders are aiming to do as soon as the detailed protocol and study has been finalised.

43. However, in projects of this nature it is not sufficient to rely on a single REC to establish a sound ethical framework. We noted that the initial plans for Biobank foresaw the need for an independent oversight body. In our report we concluded that the governance of large genetic research databases should allow for an independent body that is separate from the owners and users of the database. The composition and role of such bodies would vary, but it should provide for a mechanism to ensure that the long-term questions of access to samples and of wider benefits can be subjected to careful scrutiny.

44. We are also aware that in the case of Biobank such a body would have an important role as an independent body to hold the Biobank management to account and to maintain the necessary degree of independence from the three main Funders. We have recently heard from the Funders about the current thinking on such a body. In our view this body must command the respect of all stakeholders in the UK Biobank. We are extremely attracted to the idea that it should include individuals from different locations who have been recruited as participants into the UK Biobank. This might even include some form of elected representatives from amongst the study participants.

45. One important function of the oversight body will be to consider the wider ethical issues and the implications of any proposed nested studies working with subsets of data from UK Biobank. If these are considered in isolation, for example by an MREC, they may raise no particular concerns. However, such studies may potentially raise difficulties over feedback and re-consent. They may also contribute to a gradual "mission-creep" of the UK Biobank into potentially controversial areas of study that were not envisaged by participants.

We trust that the comments above are helpful to the Committee in considering this matter. We recognise that in places they are the product of only limited and initial consideration. If the Committee requires any additional information or clarification we will be happy to provide it. We also welcome any future conclusions and comments from the Committee that we may take into account in our future discussions of this important and necessary project.

Human Genetics Commission
November 2002

Annex A

Inside Information – balancing interests in the use of personal genetic information

In November 2000 HGC published a discussion document called *"Whose hands on your genes?"* which set out a series of questions about the storage, protection and use of personal genetic information. The use of personal genetic information in research was covered in some detail. We sought views on the general approach to consent and ownership of donated material and on the establishment of large-scale genetic databases linked to health records. We also considered the wider implications, such as the use of research databases and findings by insurers, employers and by the police force.

The consultation finished in March 2001 and we received over 250 written responses, including 86 detailed responses from organisations and individuals. We sought additional information from organisations, from insurance companies (on genetics and insurance) and also from the recently established HGC Consultative Panel of people affected by a genetic disorder. The HGC Working Group spent a considerable amount of time considering this material and discussed their draft report a number of times at the plenary Commission. The first stage was to consider the general definition and scope and to consider how, if at all, genetic information differed from other private personal information. We then sought to draw up some general principles that should cover the personal genetic information. In the light of this preparatory work, the Commission then considered the detailed issues around clinical uses, research uses, insurance and employment and forensic uses.

HGC's Inside Information report was published in May 2002. In launching the report Baroness Kennedy QC stressed the importance of the concepts embodied in the title of the report. In particular, there is a careful balance to be struck between respecting the interests of individuals whilst at the same time enabling the use of personal information for the benefit of families, the wider community and society at large. The Commission was anxious to ensure that suitable safeguards were in place and monitored. However, such safeguards should not hinder important biomedical research with unworkable restrictions.

The general principles laid down by HGC are particularly relevant to the establishment, and proper oversight, of large research databases such Biobank. The main principle of "respect for persons" is based on widely accepted international norms and which it expressed in the following terms:

> *"Respect for persons affirms the equal value, dignity and moral rights of each individual. Each individual is entitled to lead a life in which genetic characteristics will not be the basis of unjust discrimination or unfair or inhuman treatment."*

However, the report also considered that respect for the autonomy of individuals was not the only value to be taken into account. Individuals live in society, and the interests of others must be considered in the exercise of individual autonomy. We each owe certain duties as members and citizens. For example, although nobody should be compelled to participate in genetic research, the decision to participate or not should be reached in an awareness of the fact that participation may provide help to those suffering from disease.

The report highlighted circumstances in which society would seek to balance the demands of autonomy (for example confidentiality) with the interests of others. This was felt to be sufficiently important to be expressed in as the *concept of genetic solidarity and altruism* which can be summarised as follows:

> *We all share the same basic human genome, although there are individual variations which distinguish us from other people. Most of our genetic characteristics will be present in others. This sharing of our genetic constitution not only gives rise to opportunities to help others but it also highlights our common interest in the fruits of medically-based genetic research.*

This concept should be taken into account at all stages of the ethical debate over personal genetic information but is not on a par with the principle of respect for persons. That is the overarching principle from which we believe a number of secondary principles may be derived. The report was confined to the aspects that are particularly relevant to genetics. These secondary principles are:

The principle of privacy

The principle of consent

The principle of confidentiality

The principle of non-discrimination.

Copies of the report are available at www.hgc.gov.uk/insideinformation/

GOVERNMENT RESPONSE TO THE HGC'S REPORT

Inside Information: Balancing Interests in the Use of Personal Genetic Data

Baroness Helena Kennedy QC
Chair, Human Genetics Commission
Area 652,
Skipton House
80 London Road
London SE1 6LH

24 June 2003

Dear Helena,

GOVERNMENT RESPONSE TO THE HGC'S REPORT: INSIDE INFORMATION: BALANCING INTERESTS IN THE USE OF PERSONAL GENETIC DATA

When Lord Hunt and Lord Sainsbury wrote to you last July welcoming the publication of Inside Information – your report on personal genetic information – they gave an undertaking to respond in due course to your main recommendations. Their letter commented on how useful the report would be in a providing a sound basis for developing future Government policy on human genetics. We have now published the Government's White Paper on genetics *Our Inheritance, Our Future: Realising the potential of genetics in the NHS* in which you will see how many of the key aspects of your report have influenced our thinking. The White Paper has responded to some of your major recommendations, such as the new offence of non-consensual genetic testing and the need to prevent unfair discrimination on the grounds of a person's genetic characteristics. This open letter is intended to supplement the points that were addressed in the White Paper.

In some cases we indicate the current or planned Government action, in others we also make proposals for further work by the Commission. We would hope that these could be considered during discussions on your future workplan. We have divided our response below under the main chapters that you used in your report.

What is genetic information and general principles

You noted that your key principles were based on many of the recognised international instruments and declarations. As you commented, the UK has played, and continues to play, an important role in fostering this international dialogue. You recommended that the Government take steps towards signing and ratifying the Council of Europe's Convention on Human Rights and Biomedicine (paragraph 2.7). Although the Government recognises the importance of the Convention for setting out a common approach towards bioethical issues, there are several areas in which domestic policy and legal provisions are currently undergoing development following consultation. These matters will need to be resolved before the UK is in a position to consider signing and ratifying the Convention. We are, however, supportive of the work being done on a Genetics Protocol to the Convention, and we feel sure that your work on direct genetic testing will be influential in the development of this Protocol.

Clinical practice and special cases in genetics

The Government welcomes your clear summary of the complex issues and recommended best practice in counselling, the obtaining of consent and disclosure of personal genetic information. The investment and expansion in NHS genetics services that we have announced in the White Paper builds on the excellent work that has been done by the NHS genetics healthcare professionals. We intend that the new training places we have announced will enable them to meet the increasing demands for their services in an ethical and sensitive manner.

The Government has made a legislative commitment for a new offence of non-consensual testing of genetic material in the White Paper. This follows consultation on our report, *Human Bodies, Human Choices*, in July 2002 which considered new legal requirements on the obtaining of consent before tissues or organs are used for the purposes of diagnosis or research. We intend to introduce new legislation covering these issues as soon as possible.

Your report rightly considers in detail the safeguards needed to ensure the confidentiality of personal genetic information, and also the requirements to share such information in appropriate ways. The Department of Health is committed to ensuring the security and confidentiality of patient data in the NHS. There is to be a revised Code of Practice on Patient Confidentiality for NHS staff, taking account of the greater potential for sharing electronic patient records. This will be published later this year and will form part of a wider Information Governance toolkit, to guide how personal health information is shared in the NHS.

The Government is also looking at ways to secure public trust in the way that public services handle personal data. We are currently consulting on a strategy to achieve this. The proposals include a Charter or Guarantee setting out the standards for the way public services handle people's personal information, with which all public sector organisations should comply.

The Government has taken note of your concerns about the application of the Data Protection Act to medical record-keeping (4.7). We have asked the Health Records and Data Protection Review Group to examine the matter in detail. They are due to report later in 2003 and we look forward to receiving their recommendations. Finally, we note your interest in monitoring developments in the use of genetic information for prescribing purposes, for prenatal genetic testing and for national screening programmes.

Research and genetic databases

We are pleased that your report placed emphasis on the common benefits of medical genetic research and recognises the valuable partnership between the public sector and private sector in such research. You have identified a number of important principles around consent and confidentiality and we feel certain that these will be useful to research groups, research ethics committees and research funding bodies. Your report also recognised the importance that the Government places on the balancing of interests in determining what uses medical information can be put to. The Health and Social Care Act 2001 made some carefully protected changes to the accepted duty of confidentiality. We welcome your intention to work with the Patient Information Advisory Group which considers and advises the Government on applications to use identifiable medical information for research. (5.21). We hope that this will provide an opportunity to consider any particular aspects relating to genetic information.

The Government notes your recommendation to encourage compliance with best practice in research (5.37). The Department of Health published a Research Governance Framework for Health and Social Care in 2001. There will be a second edition in 2003. It brings together standards of research governance that apply to all research which relates to the responsibilities of the Secretary of State for Health – that is research concerned with the protection and promotion of public health, research undertaken in or by the Department of Health, its non-Departmental Public Bodies and the NHS, and research undertaken by or within social care services that might have an impact on the quality of those services. This includes clinical and non-clinical research, research undertaken by NHS staff using NHS resources, and research undertaken by industry, the charities, the research councils and universities within the health and social care systems. It is also offered as a model for the governance of research in other areas where poor practice could have a direct impact on the health or well-being of the public. In implementing the Framework, the Department has set a target that by March 2004 all research-active NHS care organisations comply unless there are well-documented reasons for a longer time scale.

The Medicines and Healthcare Products Regulatory Agency has consulted on draft Regulations transposing the requirements of Directive 2001/20/EC on clinical trials of human medicines. The Medicines for Human Use (Clinical Trials) Regulations 2003 will set out a new regulatory framework for clinical trials from May 2004, which will make ethical approval a legal requirement before a clinical trial involving a medicine can proceed. A new UK Ethics Committee Authority will establish, recognise and monitor ethics committees.

We have noted your detailed recommendations on the establishment and oversight of large research databases (5.44-5.45). The White Paper reaffirms the Government's support for the UK Biobank, which is being jointly funded by the Department of Health, the Medical Research Council and the Wellcome Trust. We feel certain that your report will be influential in the detailed planning of the arrangements for the operation and oversight of the database. However, you will be aware that the Funders have already agreed to an oversight body that will be separate from the hub organisation that operates the database. The Funders have also formed an Interim Advisory Group to advise on an ethics and governance framework. This Group will be requesting comments on its recommendations and you may wish to consider responding to this consultation.

Your detailed advice on aspects of the UK Biobank has also been reflected in the report of the House of Commons Science and Technology Committee (Third Report of Session 2002-03, HC132) on the work of the Medical Research Council. The Government response to that report was published on 4 June (Cm5834) and we would encourage you to consider this response in your future discussions of the UK Biobank.

In particular, your report recommended that the possibility of police access to research databases should be put beyond doubt, by legislation if necessary (5.50). We have commented in the White Paper on the current legal arrangements governing such access under the Police and Criminal Evidence Act. We are broadly satisfied that there are sufficient controls to prevent improper access. We would encourage the Commission to work with others, such as the Association of Chief Police Officers to ensure that the arrangements are sufficiently robust and properly understood by all sides. It is also important that these and other safeguards are communicated to the wider public who may be invited to participate in such research.

Insurance and employment

It is clear from your report that there are widespread concerns about the potential use to which insurance companies or employers might put personal genetic information. The Government has made clear its position on the use of genetic test results for underwriting life and health insurance. The present moratorium with the insurance industry goes some way beyond your previous interim recommendation. We welcome the constructive recommendations that you make for further work during the moratorium and your recommendations directed at the Genetics and Insurance Committee (GAIC; paragraph 7.64). We expect GAIC to take careful note of these in considering its future work and we would encourage you to work closely with them, particularly on areas that would benefit from their knowledge of the technical aspects of actuarial science and insurance underwriting.

We look forward to seeing proposals in your new workplan and to further advice that will help in the development of Government policy in time for the end of the moratorium in 2006. It will be important to work with other bodies, patient groups and professionals, including GAIC, the Association of British Insurers, the Faculty and Institute of Actuaries, the Royal Colleges, the British Society for Human Genetics and the UK Forum for Genetics and Insurance.

With respect to the recommendation for consumer information partnerships to help provide access to affordable insurance (7.50), the Government has established Consumer Support Networks to encourage better and more joined up consumer advice services. Agencies within these, for example Citizens Advice Bureaux, could signpost clients to specialist information and advice on sources of affordable insurance, such as information from the Association of British Insurers and the names of specialist insurance brokers.

The Government has taken careful note of your conclusions and recommendations about the possible use of genetic information by employers. We agree with your conclusion that at present there is no systematic use of personal genetic information in employment. You have rightly commented about the danger that individuals could be denied employment opportunities on the basis of genetic information. However, we agree that it is also appropriate to recognise that there may be important occupational health and safety benefits from appropriate use of genetic information.

The Government has accepted your recommendation to consider the general issue of 'genetic discrimination' and the appropriate means of addressing any concerns in this area (6.41). We had previously accepted a recommendation from the Human Genetics Advisory Commission for a review of genetic testing in employment by 2005. The Government is actively considering the most appropriate mechanism for this review. In the meantime, we agree with your recommendation that the HGC, with others such as the Disability Rights Commission (DRC) and the Health and Safety Commission (HSC), should continue to monitor developments (8.23). We believe that that may best be achieved by informal and flexible joint meetings, rather than by the setting up of a formal joint committee.

You also invite employers to voluntarily inform HGC of any proposals to use genetic testing for health and safety purposes (8.19). We believe that for the time being it would be appropriate for the three Commissions (DRC, HSC and HGC) to informally consider and advise on such proposals. Such consideration and advice will undoubtedly be of great value to employers, to representatives of the employees, to occupational health doctors and to the Government.

Forensic uses of genetic information

The Government has already addressed many of your recommendations on the arrangement for oversight of the UK National DNA Database and HGC has been involved in further discussions on these matters.

One of the main recommendations relates to commitments that were given by the Government during the passage of the Criminal Justice and Police Act 2001 for a review of the arrangements for the National DNA database. The independent review of the Forensic Science Service (FSS), led by Mr Robert McFarland, has taken account of recommendations that you have made in relation to the oversight arrangements and the role of the Forensic Science Service and the Association of Chief Police Officers (9.32). It is also considering the storage of the samples which are used to generate the DNA profile (9.39). The report is due to be published in late summer.

The Home Office has also consulted on a revised Code of Practice under the Police and Criminal Evidence Act. This includes guidance to police forces on the arrangements for consent to retain samples provided voluntarily for elimination purposes. The final guidance, which reflects HGC's comments, does make it clear that the consent to test the initial elimination sample and the consent to retain the profile indefinitely must be separated and clearly explained (9.26).

Your report makes a series of thoughtful recommendations aimed at encouraging a wider understanding of the importance of DNA information for criminal justice (9.48). We welcome the willingness of the Commission to work with others, particularly the ACPO National DNA Database Board, which has operational responsibility for the DNA database. We also encourage a public dialogue about future areas of research, which may be important for the detection, prevention, and deterrence of crime. In this respect, we would ask that the Commission provide advice, as requested, to the Forensic Science Service on some of the wider ethical issues around their research programme (9.45). This will complement the more detailed consideration by research ethics committees of individual research projects that may be undertaken on behalf of the FSS or the Home Office.

DNA parentage testing

The Government broadly accepts your recommendation that guidelines on the use of parentage testing in child maintenance and immigration should reflect the intrusive and sensitive nature of such information (10.20).

The Department for Work and Pensions accepts the Human Genetics Commission recommendation about the need for guidance on the use of DNA testing in Child Support Agency cases. There is clear guidance in place for Agency staff on when to offer DNA testing. This includes guidance on the need for confidentiality and consent. DNA testing is only offered by Child Support Agency staff when the alleged parent and the parent with care cannot agree about the parentage of a child for whom maintenance is sought. The Agency cannot require a person to be tested. However, if a test is refused by the non-resident parent a child maintenance assessment can still be made and enforced on the presumption that the non-resident parent is a parent of the child concerned. It is then for the Family Courts to determine parentage if the non-resident parent decides to appeal such an outcome. In practice the great majority of non-resident parents who deny parentage of a child when approached by the Child Support Agency turn out to be the child's parent.

The child support scheme applies equally to all children of separated parents and Child Support Agency staff are trained to be sensitive to the implications of using DNA tests to establish the parentage of a child.

Guidance by the Nationality Directorate of the Home Office is sensitive to concerns about confidentiality and the range of different family structures among other cultures. The Government will continue to keep such guidance under review.

We have noted your intention to hold a further review of the arrangement for private paternity testing in the light of your review of other genetic testing services offered directly to the public. We therefore look forward to further advice from HGC on the particular aspects of consent and confidentiality that are raised by DNA paternity testing and an appropriate regulatory framework. You will want to take note of the Government's commitment to introduce new legislation outlawing the non-consensual testing of genetic information. However, we are concerned that this new offence should not interfere with proper lawful access to private paternity testing. The Government accepts the view established by the Courts that it is generally in a child's best interest to know the truth about their biological origins.

We would like to thank you once again for your thoughtful and comprehensive report. We believe that it has made an important contribution to the development of appropriate safeguards to allow society to take advantages of the advances in genetic knowledge. On behalf of the Government we encourage you to continue your excellent work on the implications of advances in genetic knowledge.

JOHN REID

ANNEX E: How HGC works and how it works (role, terms of reference, methods of working, monitoring groups and working and code of practice)

Role

The Human Genetics Commission (HGC) is the UK Government's advisory body on how new developments in human genetics will impact on people and on health care. Its remit is to give Ministers strategic advice on the "big picture" of human genetics, with a particular focus on social and ethical issues.

HGC was established in 1999 following the UK Government's comprehensive review of the regulatory and advisory framework for biotechnology. Its role should also be seen in the context of other advisory and regulatory bodies in the framework for human genetics. HGC does not direct these bodies or interfere with their lines of accountability, but works with them and help form links between them. HGC reports to Health and Science Ministers and works within the context of devolution settlements for Scotland, Wales and Northern Ireland. Government policy on human genetics is generally developed on a UK basis, but responsibility for National Health Service (NHS) genetics services is the responsibility of each devolved administration.

Terms of Reference

- To analyse current and potential developments in human genetics and advise Ministers on:
  - their likely impact on human health and healthcare;
  - their social, ethical, legal and economic implications.
- To advise on strategic priorities in the delivery of genetic services by the NHS.
- To advise on strategic priorities for research.
- To develop and implement a strategy to involve and consult the public and other stakeholders and encourage debate on the development and use of human genetic technologies and advise on ways of increasing public knowledge and understanding.
- To co-ordinate and exchange information with relevant bodies in order to:
  - identify and advise on the effectiveness of existing guidance and of the regulatory and advisory framework as a whole, taking account of European and global dimensions;
  - look at the lessons learnt from individual cases requiring regulatory decision to build up a wider picture;
- To consider specific issues related to human genetics and related technologies as requested by Ministers.
- To operate in accordance with best practice for public bodies with regard to openness, transparency, accessibility, timeliness and exchange of information.

Ways of working

A constant theme and priority within our work is to actively seek input from the public and other stakeholders and this involves a variety of consultation exercises and open meetings.

We work in accordance with best practice principles on openness and transparency. We also exchange information with other bodies in the advisory and regulatory framework, including meetings at secretariat level and between chairs.

We have established sub-groups or panels which involve both Members and external participants, and which may co-opt input from individuals. We use email and telephone conferencing when this is useful, particularly for the work of the Monitoring Groups described below.

HGC may commission work from individuals or organisations on a consultancy basis.

How we organise our work

The full Commission meets around four times a year, in different parts of the country. We meet over two days – usually holding an information-gathering session, when we invite a number of people to talk to us about a particular issue, on the first day and the plenary meeting on the second.

In 2003 we set up a more flexible structure for the way the Commission carries out its work, which is shown in the diagram on page 55. We agreed to continue to focus the main areas of work in task-orientated working groups. We also identified HGC Members to lead on a number of key issues and who work with Monitoring Groups to keep a watching brief on these areas and keep them high on our agenda.

Lead Members are asked to:

- keep HGC up to date on developments and make sure the issue remains on HGC's agenda
- advise on the need for meetings of the Monitoring Group and suggest specific pieces of work as needed
- lead on liaising with other relevant organisations and co-ordinating responses to consultations

We have set up the following **Monitoring Groups**:

- Genetic Discrimination Monitoring Group, led by Bill Albert
- Horizon-Scanning Monitoring Group, led by Veronica van Heyningen
- Intellectual Property and Genetics Monitoring Group, led by Hilary Newiss
- Public Involvement Monitoring Group, led by Geoff Watts
- Research Databases Monitoring Group, led by Martin Richards

The work of the Monitoring Groups is described in the body of this report.

We felt that there was a continuting role for the HGC Business Committee and a UK-wide Genetic Services Sub-group.

The **Business Committee** continued with its existing role:

- to provide a more responsive executive structure so that HGC can react to developments quickly and involve the Membership as fully as possible.
- the Committee will have a rotating membership, and the Chair will report directly to the HGC Chair.

and also should:

- liaise with lead members between plenary meetings and continue liaison with key organisations such as Nuffield, Wellcome
- oversee external communications:
  - press office
  - website
  - newsletter/annual report
  - editorial oversight of briefing notes

In February 2003 Geoff Watts took over as Chair of the Business Committee. It has a rolling Membership and details of its meetings and Membership are on the website (www.hgc.gov.uk).

Genetic Services Sub-group

This group revised its remit in 2003, which is now:

1) To keep under review and advise the Commission of new issues and developments in the following areas, including the routine exchange of information with other relevant bodies such as the National Screening Committee, the Genetics Commissioning Advisory Group and the Human Fertilisation and Embryology Authority:
   - Strategic issues in the delivery of genetic services by the NHS and the private sector;
   - Human genetic testing services supplied direct to the public;
   - Significant new and evolving genetic tests and screening and associated technologies; and
   - Codes of practice and guidance on the ethical, social and scientific aspects of human genetic testing services and their effectiveness.
2) To make visits to see the application of genetic technologies in NHS and private genetic service facilities across the UK
3) To prepare draft documents and reports for the Commission as required and contribute to the drafting and analysis of consultation documents and responses in relation to genetic services.

Membership:

Philip Webb (Chair)
Elizabeth Anionwu
Hilary Harris
Alastair Kent
Christine Patch
Peter Sayers
John Sulston
Frances Flinter (co-optee)
Peter Harper (Wales CMO)
Patrick Morrison (NI CMO)
Stephen Singleton (England CMO)
Rosalind Skinner (Scotland CMO)

Details of the Sub-group's meetings are on the website (www.hgc.gov.uk).

Working Groups

We decided that our working groups were very good models for taking forward large pieces of work and that we would continue to set up a specific group to deal with an indivual area of work.

Working Group on Genetic Testing Services supplied Direct to the Public

This Working Group was set up to lead on our review of genetic testing services currently being offered to the UK public and was wound down with the publication of the report *Genes direct* in May 2003.

Terms of Reference

- To consider current genetic testing services provided direct to the public and available in the UK and the adequacy of present direct and indirect controls in the UK (including services offered via the Internet), EU and overseas;
- To take evidence and consider likely developments in genetic testing technology, public attitudes, the control of other comparable services in the UK and the control of testing services, including advice on such matters as lifestyle or suggested medical treatments;
- To prepare a consultation document and to consider other methods for obtaining the views of stakeholders and the public;
- To inform HGC's recommendations to Health & Science Ministers, particularly on the possible need for any changes to the advisory and regulatory framework and the provision of advice to consumers.

Members

Philip Webb (Chair)
Elizabeth Anionwu
Ruth Evans
Hilary Harris
Sandy McCall Smith
Patrick Morrison
Hilary Newiss
John Sulston
Veronica van Heyningen
Geoff Watts

Details of the Working Group's meetings are on the website (www.hgc.gov.uk).

Working Group on Genetics and Reproductive Decision Making

This Working Group was set up in May 2003 to take forward our work consider the issues around new and developing technologies associated with human reproduction and their implications for society. It superseded the earlier Scoping Group on Genetics and Reproduction.

Terms of Reference

1. To collate information, take evidence and consider past, current and future developments in genetic services related to reproduction within the current legal framework and in terms of the technology and public attitudes towards its use.
2. To examine, in particular, advances as they relate to prenatal genetic screening services, prenatal genetic diagnosis and preimplantation genetic diagnosis.

3. To work with existing bodies responsible for regulating and/or advising Government on genetics and reproduction including the National Screening Committee and the Human Fertilisation and Embryology Authority.
4. To work with HGC groups as appropriate to develop strategies for public consultation and discussion, to develop the working group's knowledge about genetic services and horizon scan in the area of genetics and reproduction.
5. To contribute to and/or respond, where appropriate, to emergent national debates about genetics services and their implications for reproductive decision making.
6. To prepare and publish a consultation document and to consider other methods for obtaining the views of stakeholders and others.
7. To identify from consultation and deliberation, sound ethical principles appropriate to genetic advances and services related to reproduction.
8. To publish a report identifying our conclusions and recommendations pertaining to the ethical principles on genetic advances and services related to reproduction and to communicate these to Health & Science Ministers.

Members

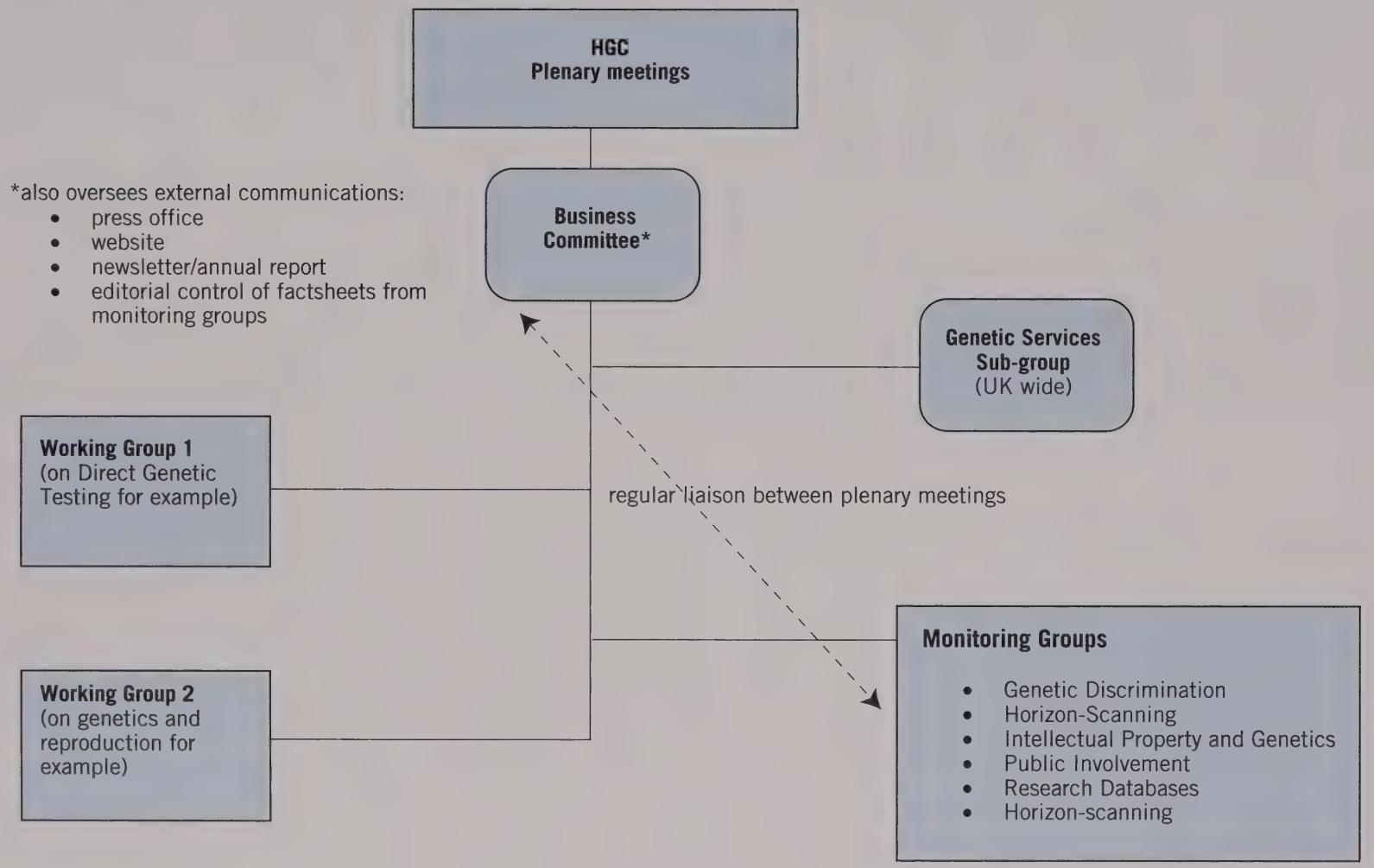
Helena Kennedy (co-Chair)
Martin Richards (co-Chair)
Bill Albert
Brenda Almond
Elizabeth Anionwu
John Burn
Heather Draper (co-opted)
Frances Flinter (co-opted)
Suzi Leather (ex officio)
Hilary Harris
John Harris
Alistair Kent
Alexander McCall Smith
Christine Patch
Peter Sayers
Martin Whittle (ex officio)

Details of the Working Group's meetings and the work of the earlier Scoping Group are on the website (www.hgc.gov.uk).

Code of Practice for Members

The HGC Code of Practice was prepared in line with Government policy on standards in public life, openness and accountability, full details are available on the HGC website: www.hgc.gov.uk. The Chair, Vice-Chair, Members and Representatives of the Chief Medical Officers (CMOs) (collectively referred to as "Members") are expected to follow it in carrying out duties associated with HGC. Co-opted members are also expected to follow the Code as it applies to the work they do on behalf of HGC.

Structure of HGC's sub-groups



Annex F: Register of HGC Members' Interests

Dr Bill Albert

Remunerated employment, office, profession, *etc*

Chair, Norfolk Coalition of Disabled People
Director, Nordat Limited, a disability awareness training organisation

Professor Brenda Almond

Remunerated employment, office, profession, *etc*

Author, editor, lecturer (occasional, free-lance).
Belle van Zuylen Visiting Professor, University of Utrecht, April-July 2003.

Miscellaneous and unremunerated interests

President of Philosophical Society of England
Vice-president of Society for Applied Philosophy
Honorary Senior Research Fellow, Social Values Research Centre, University of Hull
Overseas Member of Austrian Academy of Sciences
Honorary Fellow of Academy of Moral Sciences, Beijing University, China
Member of Societas Ethica (European Society for Ethical Research) and of European Ethics Network.

Professor Elizabeth Anionwu

Remunerated employment, office, profession, *etc*

Professor of Nursing, Head of Mary Seacole Centre for Nursing Practice, Thames Valley University

Dr Stephen Bain

Remunerated employment, office, profession, *etc*

Reader in Diabetic Medicine, University of Birmingham & Honorary Consultant Physician, Birmingham Heartlands Hospital, Birmingham, UK

Miscellaneous and unremunerated interests

Member, West Midlands Multi Research Ethics Committee
Chairman of the Pan-Birmingham Diabetes Advisory Group and the East Birmingham and Solihull Local Diabetes Services Advisory Groups.

Dr Celia Brazell

Remunerated employment, office, profession, *etc*

Director, Genetics Science and Technology, GlaxoSmithKline Research and Development

Registrable shareholdings

Aberdeen Technology Trust
Aggreko plc
The AIM Trust plc
Alliance & Leicester
Autonomy Corporation
B.A.A.
Bradford & Bingley
British Airways
Cable & Wireless
Fidelity American Fund
Fidelity UK Aggressive Unit Trust
Fidelity Special Sit Trust (1) & (2)
Gartmore UK Index fund
GlaxoSmithKline
HBOS
HSBC Holdings
Marks & Spencer
Northern Rock
Orange
Invesco Perpetual: Far Eastern Growth
Rail Track
Regus
ReNeurone
Rolls Royce
Schroders: Tokyo Fund
Scottish & Southern Energy plc
Scottish Power
Share plc
Tesco
Thus Group plc
Trafficmaster plc
Vodafone Group

Professor John Burn

Remunerated employment, office, profession, *etc*

Professor of Clinical Genetics, University of Newcastle (tenured chair, part funded by National Health Service)

Remunerated Directorships

Honorary Director, Imperial Cancer Research Fund, Clinical Cancer Genetics Network
Executive Chairman of Northgene (Identity testing) Limited, a small not-for-profit company providing a commercial paternity testing service

Miscellaneous and unremunerated interests

Director Northern Genetics Service, Newcastle NHS Hospitals Trust
Chair, Cancer Genetics Group of British Society of Human Genetics (formerly Cancer Family Study Group)
Member, Medical Advisory Board of Genetics Interest Group
Member, Ethics in Medicine Committee of Royal College of Physicians
Member, Scientific Committee of Royal College of Obstetricians & Gynaecologists

Dr Peter Harper

Remunerated employment, office, profession, *etc*

Professor of Medical Genetics, University of Wales College of Medicine, Cardiff

Dr Hilary Harris

Remunerated employment, office, profession, *etc*

General practitioner, Manchester

Professor John Harris

Remunerated employment, office, profession, *etc*

Sir David Alliance Professor of Bioethics, University of Manchester
Member, Data Safety Monitoring Board, Chiron Corporation

Baroness Helena Kennedy QC

Remunerated employment, office, profession, *etc*

Board Member, Independent Newspapers

Miscellaneous and unremunerated interests

Advisory Council Member of the Foreign Policy Centre
Member of the External Advisory Council, World Bank Institute
Patron, Charter 88
Patron, Liberty
President, Civil Liberties Trust
Trustee, KPMG Charitable Trust

Political activity

Labour Peer

Mr Alastair Kent

Remunerated employment, office, profession, *etc*

Director, Genetic Interest Group
Non-Executive Director, Cambridge City Primary Care Trust

Miscellaneous & unremunerated interests

Chair, Public Engagement Committee, NHS LifeHouse
Member, Joint Committee on Medical Genetics
Member, Association of British Insurers (ABI) Genetics Committee
Member, Genetic Commissioning Advisory Group (DH)
Member, Genetics Commissioning Group (London NHS)
Member, Orphan Medicinal Products Committee (EMEA)
Member, Progress Educational Trust Advisory Committee
Justice of the Peace, Cambridge

Ms Suzi Leather

Remunerated employment, office, profession, *etc*

Chair, Human Embryology and Fertilisation Authority

Miscellaneous and unremunerated interests
Member, Christian Socialist Movement
Individual and executive Member, National Heart Forum
Member, Child Poverty Action Group
Trustee, Food Foundation
Member, Organophosphate Information Network
Advisor to the Maternity Alliance
Honorary Senior Lectureship, Department of Epidemiology & Population Health, London School of Hygiene & Tropical Medicine
Honorary Research Fellow, Department of Biological Sciences, University of Exeter

Political activity
Labour Party Member

Professor Alexander McCall Smith
Regististrable shareholdings
GlaxoSmithKline (family)

Miscellaneous and unremunerated interests
Chair, Independent Ethics Committee, The Roslin Institute
Occasional lectures at meetings supported by pharmaceutical and other companies.

Professor Patrick Morrison
Remunerated employment, office, profession, *etc*
Consultant in Clinical Genetics, Belfast City Hospital Trust (fully funded by National Health Service)
Postgraduate Tutor and Director of the Belfast Postgraduate Centre (funded by Northern Ireland Council for Postgraduate Medical and Dental Education)

Miscellaneous and unremunerated interests
Director of Cancer Genetics, Northern Ireland Regional Genetics Service.
Member, Northern Ireland Ethics Forum

Ms Hilary Newiss
Miscellaneous and unremunerated interests
Member, BioIndustry Association (BIA)
Member, Intellectual Property Advisory Committee of BIA
Member, External Ethical Advisory Board, Pharmagene Limited

Mrs Christine Patch
Remunerated employment, office, profession, *etc*
Specialist Nurse/Genetic Counsellor Wessex Clinical Genetic Service
Postgraduate student School of Medicine University of Southampton (funded by NHS R&D training fellowship)

Professor Martin Richards
Remunerated employment, office, profession, *etc*
Professor of Family Research, Centre for Family Research, University of Cambridge
Grants, Wellcome Foundation
Previous grants, Medical Research Council and Cancer Research Campaign
Member, Wellcome Trust Medicine in Society Panel

Registrable shareholdings
CGNU Ordinary CBPO. 25 shares (formerly Norwich Union)

Miscellaneous and unremunerated interests
Member, Friends of the Earth
Member, North Cumbria Community Genetics Project Ethics Committee
Adviser to Genetics Interest Group

Mr Peter Sayers
Remunerated employment, office, profession, *etc*
Director, IDM Ltd. (Internet Design company)
Director, New Harmony Press (non-profit publishing co-operative)
Director, Accessible Globe International Ltd. (non-trading disability travel company)
Pending: Company Secretary, Salt Marketing Ltd.

Miscellaneous and unremunerated interests
Chair, Telecommunications Advisory Panel
Chair, Communications for Business
Chair, Telecommunications Industry Forum
Comparable Performance Indicators (CPI) Working Group
Member, Consumer Communications for England
Member, OFTEL's Small Business Taskforce

Dr Stephen Singleton
Remunerated employment, office, profession, *etc*
Medical Director, Northumberland and Tyne & Wear Health Authority

Dr Rosalind Skinner
Remunerated employment, office, profession, *etc*
Principal Medical Officer in the Scottish Executive Health Department
Miscellaneous and unremunerated interests
Former clinical geneticist in the University of Edinburgh

Sir John Sulston
Remunerated employment, office, profession, *etc*
Short term contract with the Wellcome Trust

Professor Veronica van Heyningen
Remunerated employment, office, profession, *etc*
Head of Cell Genetics Section, Medical Research Council, Human Genetics Unit, Edinburgh
Registrable shareholdings
GlaxoSmithKline
Unilever
Bernard Matthews (family)
Boots (family)
Diageo (family)
Elan Corp. (family)
ICI (family)
J Sainsbury (family)
Nycomed Amersham (family)
PPL Pharmaceuticals (family)
Zeneca (family)

Mr Geoff Watts
Remunerated employment, office, profession, *etc*
Journalism (writing and broadcasting), often requiring the collection of information on, the description of and the expression of opinions about topics in biology and medicine lying within the Commission's remit.
Sometimes chairs meetings and conferences, takes part in recorded discussion or acts as an occasional paid consultant to organisations which may have a commercial interest in some of the topics considered by the Commission. (No regular or continuing commitments of this kind.)

Mr Philip Webb
Remunerated employment, office, profession, *etc*
Self-employed Independent Business Advisor
Director, Hydroponic Herbs Ltd
Retired General Manager, AstraZeneca Diagnostics

Registrable shareholdings
AstraZeneca Group
Oxford Biomedica
Syngenta

Miscellaneous and unremunerated interests
Member of the Board of Trustees of the Genetic Interest Group
Chairman, Witney United Football Club

Register of Co-opted Members' Interests

Dr Heather Draper (working Group on Genetics and Reproduction)
Remunerated employment, office, profession, *etc*
Senior Lecturer, Centre for Biomedical Ethics, University of Birmingham
Occasionally paid for lectures on different aspects of medical ethics by eg hospitals, institutes of higher education and professional bodies such as the Association of Anaethetists

Miscellaneous and unrenumerated interests
Member, Unrelated Live Transplantation Regulatory Authority (ULTRA)
Member, Advisory Committee on Ethics for the Assisted Conception Unit, Birmingham Women's Hospital
Member, Ethics Advisory Board of the UK Human Tissue Bank
Member, Local Ethical Review Process, Medical School Committee, University of Birmingham

Dr Frances Flinter (Genetic Services)
Remunerated employment, office, profession, *etc*
Senior Lecturer/Honorary Consultant in Clinical Genetics, King's College London (NHS funded)

Annex G: Finance

The Human Genetics Commission is funded by the Department of Health, Office of Science and Technology and devolved administrations in Scotland, Wales and Northern Ireland.

The total spent on HGC's operation (running costs) in 2002/3 was approximately £270,000. This includes one-off additional funding of £25,000 to fund the perception audit and work on development of the HGC's website.

Staff costs – which are include a member of staff seconded from OST – and additional central support (for publications and management of the HGC website), together with the costs of the re-appointments exercise (£28,000) contributed to an estimated overall cost for HGC in the order of £475,000 in 2002/3.

The majority of this budget was spent on working in an open manner and public engagement work, with roughly:

- £95,000 spent on plenary meetings and information gathering sessions (including the cost of the Consultative Panel),
- £85,000 spent on external communications, including the Press Office and PR function and printing and publishing, and
- £30,000 spent on consultations, surveys and focus groups.

The rest of the costs were associated with running the Commission and its various activities, Sub-group and other meetings.

Fees are payable to Members at a rate of £138.66 per meeting, £172.56 per meeting for the Chair, and Members are reimbursed for all reasonable travelling expenses.

Annex H: Publications

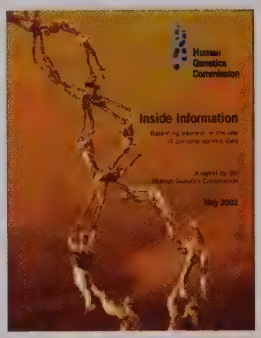
The following publications are downloadable from the HGC website (hgc.gov.uk) and in hard copy from the addresses stated.

Reports and Documents

Genes direct: Ensuring the effective oversight of genetic tests supplied directly to the public
*April 2003 (ref 31433)**

The background information and consultation responses that we considered when drawing up this report are available on our website: www.hgc.gov.uk/genesdirect/

Genetic information, public consultation
Seond Annual Report of the Human Genetics Commission
2002 (ref 30449)*



Inside Information
Balancing interests in the use of personal genetic data
*May 2002 (ref 27907)**

We drew on a wide range of evidence during our review of personal genetic information, which is available on our website: hgc.gov.uk/insideinformation/

Debating the ethical future of human genetics
First Annual Report of the Human Genetics Commission
2001 (ref 25256)*

Whose hands on your genes? November 2000 (ref 228048)*

Public attitudes to human genetic information March 2000. (ref 23992)*

*Copies of these reports can be obtained by writing to:

PO Box 777
London
SE1 6XH

Or by faxing: 01623 724524
Or by emailing: doh@prolog.uk.com

'Outcome of the Public Consultation on Preimplantation Genetic Diagnosis' (November 2001) †

HGC Public Involvement Strategy (March 2002) †

† these documents are downloadable from the website (hgc.gov.uk)

Policy Statements and consultation responses

HGC's memorandum to the HoC Science and Technology Committee on UK Biobank. November 2002†

HGC's response to the Department of Health/Welsh Assembly consultation 'Human Bodies, Human Choices'. October 2002†

HGC comments to inform the Government response to the House of Lords report on Genetic Databases. June 2001†

HGC comments to inform Government response to the House of Commons report on Genetics and Insurance. June 2001†

HGC's interim recommendations on genetic testing and insurance. May 2001†

HGC's response to HFEA on the consultation on PGD. March 2001†

† these documents are downloadable from the website (hgc.gov.uk)

Newsletter

Copies of HGC News are downloadable from the website: hgc.gov.uk/news/

You can resgister online to receive future editions or write to us to to receive copies by post

Also available on the website

HGC press notices

HGC meeting papers

Human
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It is also on our website on http://www.hgc.gov.uk/